NHS England

SCHEDULE 2 – THE SERVICES

A. Service Specifications

1. Service name	• Service name **Clinical genomic services (adults and children)** • Service sub-heading **Regional Clinical Genomic Services**
2. Service specification number	• E01/S/a
3. Date published	• TBC
4. Accountable Commissioner	• NHS England Genomics Unit
5. Population and/or geography to be served	
5.1. Population covered	

5.1.1. NHS Clinical Genomic Services (NHS CGSs) (previously called Clinical Genetics) 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. The service is responsible for the application and interpretation of genomic and other diagnostic testing and, subject to an appropriate referral, the screening and management of at-risk family members.

5.1.2. The unit of responsibility in clinical genomics is to the referred individual who presents for diagnosis, risk estimation and/or treatment. There is an acknowledged responsibility to other at risk/affected family members, whereby the referred individual is provided information to share within the family unit. This will ensure that affected and 'at risk but clinically well' relatives can be managed appropriately, offering the opportunity for diagnostic, predictive and carrier testing, screening, early intervention and prenatal/preconception genetic counselling.

Rare Disease

5.1.3. The UK Rare Disease Framework (January 2021) defines a disease or condition as rare if it affects less than 1 in 2,000 people in the population. Often rare diseases are chronic and life threatening. It is estimated that there are over 7,000 distinct rare diseases and that at least 80% have a genetic origin. Together they impact on the lives of approximately 6-8% of the population, over 3 million people in the UK. This equates to approximately 15,000 to 20,000 new diagnoses each year (Based on Office of National Statistic Population Forecast for 2021 (2019).

5.1.4. *A correct diagnosis can end what can be, for many, a lengthy 'diagnostic odyssey'. The complex nature of many rare conditions means that patients may undergo*

multiple referrals, inconclusive tests, and sometimes incorrect diagnoses before a final diagnosis is reached, during which time their condition may deteriorate, as well as having a negative impact on their mental health. Delayed diagnosis may also mean missing the window of opportunity for certain treatments, such as gene therapies, resulting in poorer outcomes. Alongside great personal cost, research from 2018 estimated that over a 10-year period the 'diagnostic odyssey' for rare diseases costs NHS England in excess of £3.4 billion (England Rare Disease Action Plan, 2022).

Inherited Cancer

5.1.5. A gene change found in every cell in the body is called a germline variant. Germline variants are nearly always inherited from a parent. Cancers caused by germline variants are called inherited cancers and account for 5-10% of all cancers.

5.1.6. A total of 305,683 service users were diagnosed with cancer in 2017 across England (Office of National Statistics (ONS). As 5-10% of cancers arise in individuals with a genetic predisposition, approximately 16,000 to 33,000 new incidences of inherited cancers each year. These service users have a significantly increased risk of developing certain types of tumour.

5.1.7. Genomic tests of tumour tissue, and other samples such as saliva and blood, for service users with cancer may identify inherited genetic variants that can indicate heritable risks to other family members. Specialist Cancer Genetics Teams within the NHS CGS support colleagues in oncology by providing advice to affected individuals along with genomic testing and counselling for at risk family members.

Sporadic (non-familial) cancer

5.1.8. Most cancers are the result of acquired, or *somatic*, mutations (these are sporadic genetic variants) that occur in a cell. Somatic mutations are often caused by environmental and lifestyle factors. Genomic testing of tumour tissue is designed to detect somatic mutations and aims to guide personalised cancer care. Somatic genomic testing does not normally detect germline genetic variants, but in some cases (e.g. colorectal or endometrial cancer associated with Lynch syndrome) can reveal the risk of a familial variant predisposing to inherited cancer.

5.1.9. Many oncologists (cancer specialists) regularly request somatic genomic testing as a routine diagnostic and to understand the information it provides and how it can influence service users' care. It is directly linked to their prescribing/treatment role. However, there are occasions when variants detected in tumour tissue are complex or previously unseen. In these circumstances, the specialist Cancer Genetics Teams within the NHS CGS will support oncologists, providing them with the necessary advice and guidance they require, to enable them to effectively integrate the genetic results into patients' care.

5.1.10. The NHS Long Term Plan aspires to offering more extensive genomic testing to patients who are newly diagnosed with cancers so that, by 2023, over 100,000 people a year will access cancer genomic testing. This growth in cancer genomic testing is likely to increase the range of cancer specialist services requesting genomic tests. As a result, oncologists may require greater multi-disciplinary support until testing pathways become more established. This support will be gained through the NHS Genomic Laboratory Hub cancer medical teams to advise on interpretation of the results and the NHS CGS Cancer Genetics Teams to provide the necessary training and upskilling to understand how the results may impact on service user care.

Other service user groups

5.1.11. Some genomic conditions are more common than the definition of a rare disease and are caused by a single change in a chromosome or gene. These more common genetic conditions affect service users with some familial cancers (e.g. hereditary breast cancer and ovarian cancer and Lynch Syndrome), familial hypercholesterolaemia and hypertrophic cardiomyopathy. These service users

may be cared for in other hospital specialities or by primary care but will require the specialist input from NHS CGSs.

Future Demand

5.1.12. Increasingly, new genetic conditions are being identified as technologies and treatments are developed. These developments are likely to increase future demand for services.

5.1.13. Through the mainstreaming of genomics into other clinical specialties (see Para. 7.1.18), the number of service users referred to the NHS CGS to access testing and diagnosis is expected to reduce as the clinical specialties become confident to take on this role. However, the increase in genomic awareness and testing in mainstream services is expected to increase the number of service users diagnosed with a rare disease or cancer/risk of cancer with a genetic predisposition, which will lead to a higher number of service users with complex disorders referred to the NHS CGS who require long term management and increased clinical and psychosocial input.

5.2. Minimum population size

Not used

6. Service aims and outcomes

6.1. Service aims

6.1.1. To provide a comprehensive service for the diagnosis, management and support of service users with complex, rare and/or inherited, often undiagnosed genetic conditions (including those at risk of hereditary cancers) and malformation syndromes and their families to promote improved clinical management and quality of life for those affected. Accurate diagnosis is dependent on clinical assessment and combining laboratory analyses with expert input from health professionals experienced in the clinical interpretation of genomic data so the Provider will take a networked approach to service delivery alongside NHS Genomic Medicine Service (NHS GMS) Alliances NHS Genomic Laboratory Hubs (NHS GLHs) and other clinical and laboratory specialties.

6.1.2. Service users and families affected by or at risk of a genetic disorder will be supported to understand their condition, its implications, and their options in relation to reproduction, screening, prevention and clinical management.

6.1.3. Care for service users and their families will be provided in a co-ordinated way; the Provider working together with other parts of the health and care system to enable involvement of everyone contributing to the patient's care to avoid duplication and achieve shared outcomes. Co-ordination will be family-centred and include service users' medical, psychosocial, educational, and vocational needs irrespective of diagnosis, service user circumstances and/or geographical location.

6.1.4. Access to the service will be equitable across the entire eligible catchment population and there will be opportunities for all service users and their families to access relevant genomic testing, in line with their clinical need and the National Genomic Test Directory, to support accurate, rapid diagnosis and implementation of new treatment approaches where appropriate.

6.1.5. Barriers to accessing the NHS CGS, genomic testing and clinical research will be reduced. In partnership with the NHS GMS Alliance, the Provider will improve engagement with communities that have historically not been reached, increasing awareness of the NHS CGS across other healthcare providers and the local population.

6.1.6. In line with their local demographic profile the Provider will increase the diversity of genomic testing and data, for example through their contribution to Genomics England's 'Diverse Data' initiative to increase representation of minority ethnic groups in the genomic research programme which aims to tackle health inequalities by recruiting at least 15% of people from ethnic minority backgrounds

to take part in research programmes thereby reducing the historical bias towards populations of European ancestry.

6.1.7. The Provider will contribute to implementation of the England Rare Disease Action Plan, in particular the reduction in diagnostic odyssey for service users with rare diseases, helping them to get a final diagnosis faster. In addition, the Provider will ensure the necessary support to service users with rare diseases to improve their quality of life through the development of pathways that provide wider support and co-ordinated care with hospital specialties and post diagnostic care, and improved access to advice and supportive resources. This will include co-ordinated pathways for access to specialist care, treatment, drugs, social care, mental health and special educational support.

6.1.8. The Provider will have a Cancer Genetics Team within its NHS CGS that will increase the utilisation of genomic data in cancer care. Specifically, the Provider will have pathways in place that support an increase in germline testing to identify those people who are more susceptible to cancer and longer term support for those service users with a genetic variant that require annual screening and/or are considering risk reducing surgery. The Cancer Genetics Team will also support oncologists and cancer services as they extend somatic genomic testing of cancer tissue, providing them with the necessary advice, guidance and training needed to embed somatic genomic testing into personalised care for service users with cancer.

To Note: In Year One, following publication of this Service Specification, the Provider will contribute to the development and implementation of a nationally consistent methodology to measure the impact that the service has on patient outcomes. It's acknowledged that measuring standard health-related outcomes are not always appropriate to those that receive care from NHS CGSs so outcomes are likely to be based on improved psychological resilience and empowerment to make informed decisions about their or their child(ren)'s future.

6.2. Outcomes

NHS Outcomes Framework Domains & Indicators

Domain 1	Preventing people from dying prematurely
Domain 2	Enhancing quality of life for people with long-term conditions
Domain 3	Helping people to recover from episodes of ill-health or following injury
Domain 4	Ensuring people have a positive experience of care
Domain 5	Treating and caring for people in a safe environment and protecting them from avoidable harm

Service defined outcomes/outputs

No	Indicator	Numerator	Denominator	Data source	Domains	CQC Key Question
1	Proportion of cases discussed at Genomics-led MDTs where clinical outcome of cases recorded and actioned	No. with outcome recorded in patient record	All service users discussed at MDT	Local Audit	4	Effective
2	Proportion of cases undergoing Whole Genome Sequencing (WGS) for a rare disease (Clinical Indication R27) and where outcome is recorded (noting where MDT discussion is required) resulting in a definitive diagnosis or change in clinical management	No. with outcome recorded in patient record	Number of cases undergoing WGS for rare disease (Clinical Indication R27)	Local Audit	4	Effective
3	Proportion of cases undergoing fetal exome analysis where outcome is recorded and confirmed or changed clinical diagnosis or management	No. with outcome recorded in patient record	Number of cases undergoing fetal exome analysis (Clinical Indication R21)	Local Audit	4	Effective
4	Proportion of neonates undergoing rapid exome analysis where outcome is recorded and confirmed or changed clinical diagnosis or management	No. with outcome recorded in patient record	Number of neonates undergoing rapid exome analysis (Clinical Indication R14)	Local Audit	4	Effective
5	Proportion of women with a pathogenic variant in *BRCA1*, *BRCA2* or *PALB2* at high risk of breast-ovarian cancer who have had risk and benefits discussion of risk reducing options	Number who have had risk and benefits discussion regarding risk reducing options	Number of mutation positive women at high risk of breast-ovarian cancer	Local Audit	2,3,4	Effective

7. Service description

7.1. Service Model

National Genomic Medicine Service Structure

7.1.1. The seventeen regional NHS CGSs across England will be embedded within the national NHS GMS (See Annex A). The NHS GMS consists of the NHS CGSs, seven NHS GLHs who are responsible for the delivery and performance of the genomic testing within their geographical region and seven NHS GMS Alliances which are aligned to the NHS GLH geographical footprints (See Figure 1).

7.1.2. Each Provider of NHS CGSs will be accountable to its host trust and, through them, to NHS England's Genomics Unit as commissioner of the service. The Provider, and its host Trust, will be responsible all aspects of performance of the service in relation to its geographical area, the local population, and its contribution to the NHS GMS priorities. Performance will be monitored and managed by NHS England.

7.1.3. The NHS GMS Alliances oversee and co-ordinate the embedding of genomics into routine care locally and across the whole population the NHS serves in England by bringing together the vital multi-disciplinary clinical leadership and other operational and digital functions that are necessary to make this possible.

7.1.4. The seventeen regional NHS CGSs are each responsible for a defined geographical area. Each of these services will take a networked approach for delivery of this service specification. Networks will be formed across the seven NHS GLHs and NHS GMS Alliance geographical footprints to optimise access to and provision of NHS CGSs. These seven regional networks will also work as part of the NHS GMS national network to share best clinical practice, support workforce development, education and training, research, develop guidance, set direction for the NHS GMS as a whole and ensure equitable access for service users and their families, recommending priorities for service improvement when identified.

7.1.5. The Provider will work to understand the needs of the local population and work with their NHS GMS Alliance and other stakeholders to address identified unmet need.

Delivery of Clinical Genomic Services

7.1.6. The Provider will have in place a mixed model of service delivery, including face to face consultations and virtual arrangements for video and telephone consultations and app-based platforms as deemed appropriate to meet local service user needs.

7.1.7. Face to face outpatient consultations will be provided through a Hub and Spoke Model, with a single Provider responsible for delivering the NHS CGS from outpatient facilities within the host Trust and via outreach within district general hospitals or other suitable settings across the geographical catchment area of the service. Where clinically appropriate and agreed to by the service user, the Provider will opt for a 'virtual' approach to care to minimise the burden of travel on both service users and staff.

7.1.8. The Provider will facilitate co-ordination and health surveillance for complex, multi-system genetic disorders, including for those with a genetic predisposition to cancer.

7.1.9. The Provider will undertake inpatient assessments where clinically appropriate. The Provider will also have provision in place to meet service user needs in urgent clinical scenarios, ensuring patient assessment and management can be met in clinically appropriate timescales.

7.1.10. The Provider will undertake joint consultations with other specialties to improve clinical pathways, reduce time to diagnosis, facilitate ongoing management and reduce the burden on service users.

7.1.11. Clinical care including advice, diagnosis and clinical management may be provided by NHS CGS health professionals without direct patient contact via

letters/email/telephone conversations and through other methods if clinically appropriate. Clinicians will review genomic and phenotype data and advise the managing clinician directly or as part of a multi-disciplinary team meeting.

Multi-Disciplinary Approach

7.1.12. The Provider will work with its NHS GLH to ensure the delivery of effective clinical input for the selection, interpretation and reporting of clinical genomic tests and provide specialist input into clinical genomic multi-disciplinary team meetings (MDTs) and Genomic Test Advisory Boards (GTABs).

7.1.13. The NHS CGS will participate in clinical MDTs, either face to face or virtually, with clinicians from a variety of specialties across its geographical region to facilitate diagnosis and enable appropriate access to genomic testing where indicated.

7.1.14. The Provider will ensure that a co-ordinated approach is taken, working together with other parts of the health and care system, to enable involvement of everyone involved in the service user's care to avoid duplication and achieve shared outcomes. As such, the Provider will ensure that processes are in place to provide written, verbal, virtual and electronic advice to colleagues in other medical specialties and in primary care to optimise the care of their patients and the need for referral to the NHS CGS.

7.1.15. The Cancer Genetics Team, which will be an integral part of the NHS CGS, will work in a co-ordinated way with cancer specialities to implement pathways for service users who have a potential inherited pathological variant identified in tumour tissue to appropriate inherited cancer MDTs, counselling, genomic testing and interpretation of clinical results. The Cancer Genetics Team will also support oncologists and cancer services as they extend somatic genomic cancer testing, providing them with the necessary advice, guidance and training needed to embed somatic genomic testing into personalised care for service users with cancer.

7.1.16. The Provider will work collaboratively with other elements of the NHS GMS to establish an operational plan for provision of the appropriate workforce and expertise required to support the rare disease and inherited cancer MDTs and to jointly develop the MDT tools required to facilitate effective MDT delivery.

7.1.17. Where the Provider has specific expertise in a particular specialist area of genomics that is not widely available within the NHS GMS, contribution to genomic MDTs will be made on a national level.

Genomics in Mainstream Clinical Pathways

7.1.18. The Provider, working with the NHS GLH and NHS GMS Alliance, will play a key role in ensuring that genomics medicine is mainstreamed (i.e. primarily delivered in hospital clinical specialties and primary care). Joint working will support an increase in confidence within mainstream services to directly apply genomic testing in line with the National Genomic Test Directory and understand how results can impact on service user management, particularly in relation to pharmacogenomics and personalised care.

7.1.19. The Provider, in partnership with its respective NHS GLH, NHS GMS Alliance and Health Education England, will support the training and education of new and existing healthcare science workforce and the broader workforce to make best use across the NHS of expertise in genomic technologies and genomic application for service user benefit.

7.1.20. The Provider will work alongside the NHS GMS Alliance to engage with hospital specialties to develop pathways where they do not already exist, and maintain/further develop pathways where they are already established, and embed genomic testing and/or advice for all service users with genetic conditions and where genomic testing is provided as part of cancer care. This support may take the form of multidisciplinary clinics, MDTs, joint agreement of referral pathways to ensure patients with complex needs are referred to the NHS CGS and/or other appropriate clinical services.

7.1.21. The Provider will have facilities in place to provide real-time advice, navigation and support for mainstream and oncology clinical colleagues to enable the uptake and embedding of genomic medicine through appropriate pathways.

7.1.22. The Provider will support colleagues in cancer teams in relation to the management of service users affected by inherited cancers and facilitate genomic testing and counselling for at risk family members. The Provider will also provide advice as required to support tumour specific genomic testing and interpretation.

7.1.23. The Provider will support mainstream hospital specialties with clinical care of service users with less complex needs through the provision of expert advice by phone, video link or e-mail. The Provider will directly support hospital specialties until genomic-informed care is consistent, pathways are embedded and the delivery of services for more common genetic conditions can be managed effectively through mainstream hospital specialities.

7.1.24. The Provider will ensure that the clinical interpretation of the outcomes of genomic testing is undertaken by staff with experience in interpreting the data, supporting mainstream services with the interpretation of results as necessary.

7.1.25. The Provider will work alongside the NHS GMS and other clinical services to determine how new aspects of genomic data, including pharmacogenomics, are integrated into routine clinical care where appropriate and where there is evidence of service user benefit.

7.1.26. The Provider's Cancer Genetics Team, in collaboration with its respective NHS GLH and NHS GMS Alliance, will support education in primary care and secondary care to enable those services to identify, triage and reassure those service users not at increased risk, facilitate screening for those at moderate risk and refer those at higher familial risk who require expert NHS CGS input.

7.1.27. The Provider will support implementation and delivery of NHS GMS Alliance transformation programmes.

7.1.28. Polygenic Risk Scores indicate the genetic contribution to common conditions such as hypertension and breast cancer, and can be used to determine disease aetiology, define risks to relatives and select optimum management. The Provider will work alongside colleagues in clinical specialties across its geography to develop and implement a plan for the incorporation of Polygenic Risk Scores into routine clinical care where relevant.

Research

7.1.29. The Provider will work with their respective NHS GMS Alliances, NHS GLHs and the wider NHS GMS to undertake research and quality improvement schemes to determine the evidence for changes to clinical practice to ensure optimal patient care and experience and as part of the NHS Genomic Medicine Service Research Collaborative and NIHR.

7.1.30. The Provider will demonstrate translation of research into clinical practice and will support the adoption of cutting-edge genomic technologies into healthcare.

7.1.31. The Provider will support future national genomics projects and initiatives, including Genome UK: the future of healthcare, the Rare Disease Action Plan and others as approved by the NHS England Genomics Unit with partners such as, Genomics England, and other funded national and international projects for patient benefit.

7.2. Pathways

Overall patient pathway

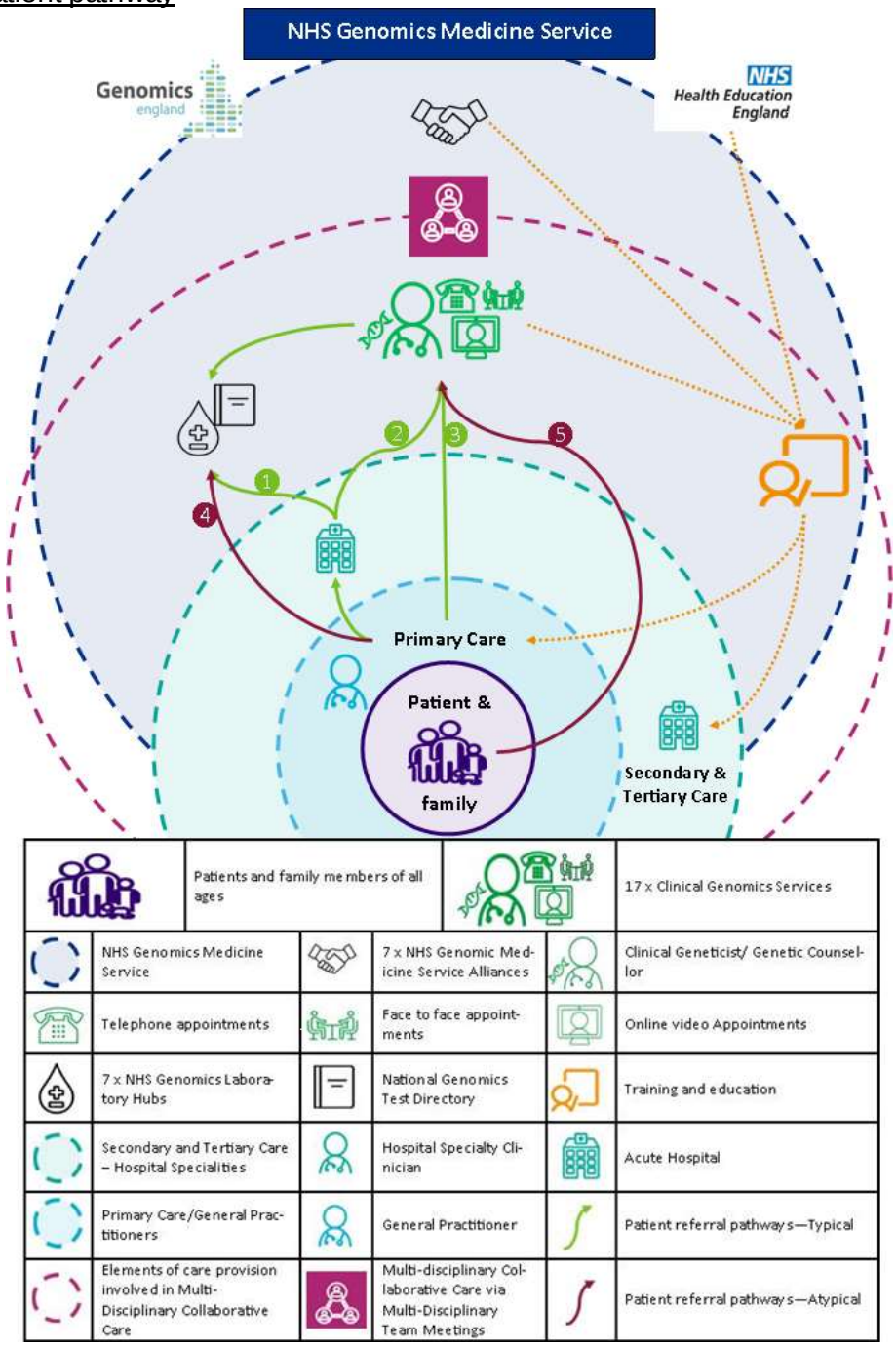


Figure 1: NHS Genomic Medicine Service Patient Pathways

Service User Pathway	Service User Pathway Description
1	The service user and/or their family is referred by their General Practitioner to a secondary/tertiary hospital speciality. The speciality clinician understands how genomics might inform the diagnosis, treatment options and management of their patient and so requests genomic testing, in line with the National Genomic Test Directory, directly from their local NHS Genomics Laboratory Hub. The speciality

	clinician receives support with interpretation of the genomic testing result from Clinical Scientists within the NHS Genomics Laboratory Hub and/or Clinical Geneticist/Genetic Counsellor (Genomic Counsellor[1]) within the NHS CGS via Multi-Disciplinary Team Meetings. The speciality clinician may then manage the service user's ongoing care, with or without direct support from the NHS CGS, or refer them to the NHS CGS with a confirmed diagnosis for ongoing management, including psychosocial support, if their condition is especially rare, complex or there is specific expertise within the NHS CGS.
2	The service user and/or their family member is referred by their General Practitioner to a secondary/tertiary hospital speciality. The speciality clinician requires specialist input from the NHS CGS because the service user's condition is complex and meets the eligibility criteria for referral. The Clinical Geneticist/Genetic Counsellor will undertake diagnostic evaluation (history taking/clinical examination and diagnostic testing, including genomic testing), manage the service user's psychosocial needs, requests for genomic testing, subsequent interpretation of results for the service user and ongoing support and management, continuing to provide co-ordinated care and liaising with other clinicians involved in the service users overall pathway.
3	The service user and/or their family member is referred directly to the NHS CGS by the General Practitioner. The Clinical Geneticist/Genetic Counsellor undertakes diagnostic evaluation, interpretation of results for the service user, manages the psychosocial support of the service user and family, and provides co-ordinated care and liaises with other clinicians providing care.
4	In some circumstances, and when deemed clinically appropriate, in line with the National Genomic Test Directory and in accordance with the acceptable Requesting Specialties set out in the National Genomic Test Directory Testing Criteria, a General Practitioner may directly request genomic testing from the NHS Genomic Laboratory Hub. The General Practitioner may then manage the service user's ongoing care, with or without direct support from the NHS CGS, or refer them with a confirmed diagnosis to the NHS CGS for ongoing management, including psychosocial support, if their condition is complex and meets referral eligibility.

[1] The term 'Genetic Counsellor' is an accepted professional title. 'Genetics' scrutinises the functioning and composition of a single gene whereas 'genomics' addresses all genes and their interrelationships to identify their combined influence.

NHS England would like to propose that work is undertaken jointly with Association of Genetic Nurses and Counsellors members and the professional registration service to update the professional registration to 'Genomic Counsellor'

NHS England believes that the term 'Genomic Counsellor' better reflects the contribution made by the profession to the NHS Genomic Medicine Service's objective to understand more about complex diseases caused by multiple genes and environmental factors, leading to earlier diagnoses, interventions and targeted treatments.

NHS England will explore this proposal further as part of the Public Consultation associated with this specification. The ongoing descriptor will be informed by the outcome of the consultation feedback.

5	In some circumstances, service users and/or their family members may feel that they would benefit from genomic testing but have exhausted attempts to access the specialist NHS CGS via their General Practitioner and/or speciality clinician. Service users and/or their family members may contact the NHS CGS directly. The Clinical Geneticist/Genetic Counsellor will assess the individual's clinical appropriateness for referral to the NHS CGS. When a self-referral is deemed not clinically appropriate, the Provider will have processes in place to provide written and/or telephone advice for service users, including signposting to a more appropriate service and provision of general advice and appropriate resources. If deemed clinically appropriate, the NHS CGS will arrange an appointment with the service user and undertake diagnostic evaluation, interpretation of results, manage psychosocial support of the service user and family, provide co-ordinated care and liaise with other clinicians providing care.

Specialised patient pathway

Referral and Triage

7.2.1. The Provider will accept referrals for service users for the diagnosis, investigation, management, risk assessment and psychosocial support of patients with complex, rare and/or inherited, undiagnosed conditions (including those at risk of hereditary cancers) from primary, secondary and tertiary care in line with nationally consistent eligibility criteria and any local eligibility criteria relevant to specific local expertise and specialisms.

7.2.2. The Provider will accept referrals for relatives of service users when they are at significant risk of a genetic condition or for appropriate risk assessment, genomic testing, organisation of surveillance and time limited psychosocial support.

7.2.3. Patients with ultra-rare or complex multi-system conditions may be referred across the geographical boundaries of individual services to specialist NHS CGSs or to NHS Highly Specialised Services (HSS) to access specialist expertise, surveillance, management and treatment.

7.2.4. The Provider will accept referrals for unaffected at-risk service users considering predictive and/or pre-symptomatic genetic testing.

7.2.5. Where access to referral is not achieved through typical routes, individuals may self-refer to the service. The Provider will ensure that information in relation to eligibility and the process for patients to refer themselves to the service is publicly available. The Provider will assess the service user's eligibility, providing them with relevant guidance and/or offer an appointment.

7.2.6. The Provider will have referral guidelines and triage processes in place to determine how referrals will be managed, the most appropriate staff to provide care and modality of clinical contact (face to face, telephone, video clinic or written advice to the referring clinician). Guidelines and processes will allow the necessary flexibility to meet local need and reduce barriers to access.

7.2.7. The Provider will have processes in place to provide written and/or telephone advice and management recommendations for service users that are not offered an appointment due to ineligibility, including signposting to a more appropriate service and provision of general advice and appropriate resources.

Service User and family care

7.2.8. A clinic appointment with a Consultant in Clinical Genetics (or medical staff under consultant supervision) is organised if the patient requires a detailed medical clinical assessment, including a clinical examination to allow investigations, diagnosis, management options and consequent screening of at risk family members and pre-natal options to be discussed.

7.2.9. A clinic appointment with a Genetic Counsellor will be offered for referral indications and patient groups where a clinical assessment or examination is not required (although clinical measurements may be taken), so that testing and management options and consequent screening of at risk family members and pre-natal options can be discussed and supported through the utilisation of counselling frameworks and skills.

7.2.10. The Provider will facilitate the ordering of relevant diagnostic testing, including genomic testing in line with the National Genomic Test Directory.

7.2.11. Where ongoing health surveillance is required for complex multi-system or pre-symptomatic conditions, the Provider will co-ordinate service user management with relevant hospital specialties to reduce duplication and achieve shared goals for service users and their families.

7.2.12. The Provider will review service users in whom a genomic diagnosis is likely but genomic testing is negative or requires additional interpretation, and where further input will either enable clinically focussed reanalysis of data or expert clinical evaluation to improve the likelihood of a diagnosis, enabling risk prediction, management and treatment.

7.2.13. Where relevant treatments for specific rare or complex genetic conditions become available, the Provider will have local arrangements in place for these to be prescribed within the NHS CGS.

7.2.14. The Provider will follow-up and support service users and families with rare genetic disorders where clinical expertise and management is not available through other services, enabling access to clinical trials and supporting the development of new specialist services by other clinical specialities.

7.2.15. The Provider will contribute to MDT clinics with other hospital specialists for service users with complex and/or multi-system disorders where this best meets the needs of individuals.

7.2.16. The Provider will advise service users of their prenatal options and facilitate access to Preimplantation Genetic Diagnosis services.

7.2.17. The Provider will ensure that access to specialist Clinical Psychology is available for service users, either through direct recruitment of clinical psychologists into the Provider's workforce or through pathways to specialist local mental health service provision.

Shared care arrangements

See Section 7.1: Service Model - Genomics in Mainstream Clinical Pathways

7.3. Clinical Networks

All Providers will be required to participate in a networked model of care to enable services to be delivered as part of a co-ordinated, combined whole system approach.

Clinical Network/ODN	Link to 'published' network/ODN specification
NHS GMS Alliances	[Not yet published – link to be included when available]
Rare Disease Collaborative Networks	Rare Disease Collaborative Networks
Cancer Alliances	Detailed here
National institute for Health Research Clinical Research network	Detailed here
NHS Genomic Medicine Service Research Collaborative	Detailed here

Genomics England Clinical Interpretation Partnership	Detailed here

7.4. Essential Staff Groups

7.4.1. The Provider will have roles within their workforce that enable them to work effectively and to manage required changes to the care model and contribution to the wider NHS GMS. These are likely to include:

- Clinical Geneticists (Including Clinical Geneticists with Inherited Cancer expertise).
- Genetic Counsellors.
- Adequate mix of other staff groups to meet population needs and demand for the service including General Practitioners with Special Interest, Family History Co-ordinators, Genomic Associates, Genomic Assistants, Genomic Practitioners and Specialist Nurses.
- Administrative staff.
- Specialist Clinical Psychologists (or direct pathways to specialist psychology in local Mental Health Services).

7.4.2. The Provider will host clinical geneticist, genetic counselling, psychology, nursing and any other relevant trainees to support building the future workforce required to meet increasing demand for services.

7.4.3. At a minimum, the Provider will have a Lead Clinical Geneticist and Lead Genetic Counsellor with lines of reporting to the relevant Directorate Lead. These roles will ensure delivery of service priorities and include networking and collaborative working with the NHS GMS to ensure aligned decision making, representing the Provider at meetings and working groups that are part of the governance and organisational structure of the NHS GMS.

7.4.4. The Provider's Genetic Counsellors will be key to the mainstreaming agenda and their role will include the embedding of pathways, up-skilling mainstream clinicians and providing real-time advice and guidance to enable a seamless service user pathway across specialties will be formalised, implemented and embedded.

7.4.5. Where appropriate to enhance a networked approach to service delivery and to support the mainstreaming agenda, the Provider will embed clinical genomic staff into mainstream clinical specialties. These staff will be wholly supported by the Provider, enabling ongoing links and engagement with the NHS CGS.

7.4.6. The Provider will work collaboratively within the NHS GMS, and with Health Education England, professional bodies, local deaneries (organisations responsible for post-graduate medical training) to create, maintain and provide workforce development plans that reflect the requirements to meet ongoing local demand and population needs.

7.5. Essential equipment and/or facilities

Not applicable

7.6. Inter-dependant Service Components – Links with other NHS services

The table below lists services that NHS CGS is reliant on; or are/will be reliant on the NHS CGS for genomic expertise.

Interdependent Service	**Relevant Service Specification/Standards**	**Proximity to service** (not applicable/co-located/same town/city)
NHS Genomic Laboratory Hubs	NHS England » Genomic Laboratory Hubs	Not applicable

Neonatal Critical Care	https://www.england.nhs.uk/wp-content/uploads/2018/08/Neonatal-critical-care.pdf	Across the geography served
Paediatric Intensive Care	https://www.england.nhs.uk/wp-content/uploads/2018/08/Paediatric-intensive-care.pdf	Across the geography served
Adult Critical Care	https://www.england.nhs.uk/publication/adult-critical-care-services/	Across the geography served
Oncology/ Cancer Centres	See NHS England » Service specifications for cancer related service specifications	Across the geography served
Fetal medicine	https://www.england.nhs.uk/wp-content/uploads/2018/08/Fetal-medicine.pdf	Across the geography served
Neurology	https://www.england.nhs.uk/wp-content/uploads/2018/08/Neurosciences-specialised-neurology-adult.pdf	Across the geography served
Cardiology	https://www.england.nhs.uk/publication/cardiology-inherited-cardiac-conditions-all-ages/	Not applicable
Renal		Not applicable
Primary Care		Not applicable
Clinical Psychology		Not applicable
Pathology		Same hospital
Paediatric wards		Across the geography served
Paediatric Medicine	See NHS England » Service specifications for paediatric medicine related service specifications	Not applicable
Prescribed Specialised Services for service users with genetically predisposed conditions	See relevant sections within the prescribed-specialised-services-manual.pdf (england.nhs.uk)	Not applicable

7.7. Additional requirements

7.7.1. The Provider will support delivery of pilot approaches for patients with undiagnosed rare conditions as per the England Rare Disease Action Plan, and other pilot developments introduced within the NHS GMS.

7.8. Commissioned providers

7.8.1. See Annexe A

7.9. Links to other key documents

Please refer to the *Prescribed Specialised Services Manual* for information on how the services covered by this specification are commissioned, contracted and paid for (as described in the Identification Rules).

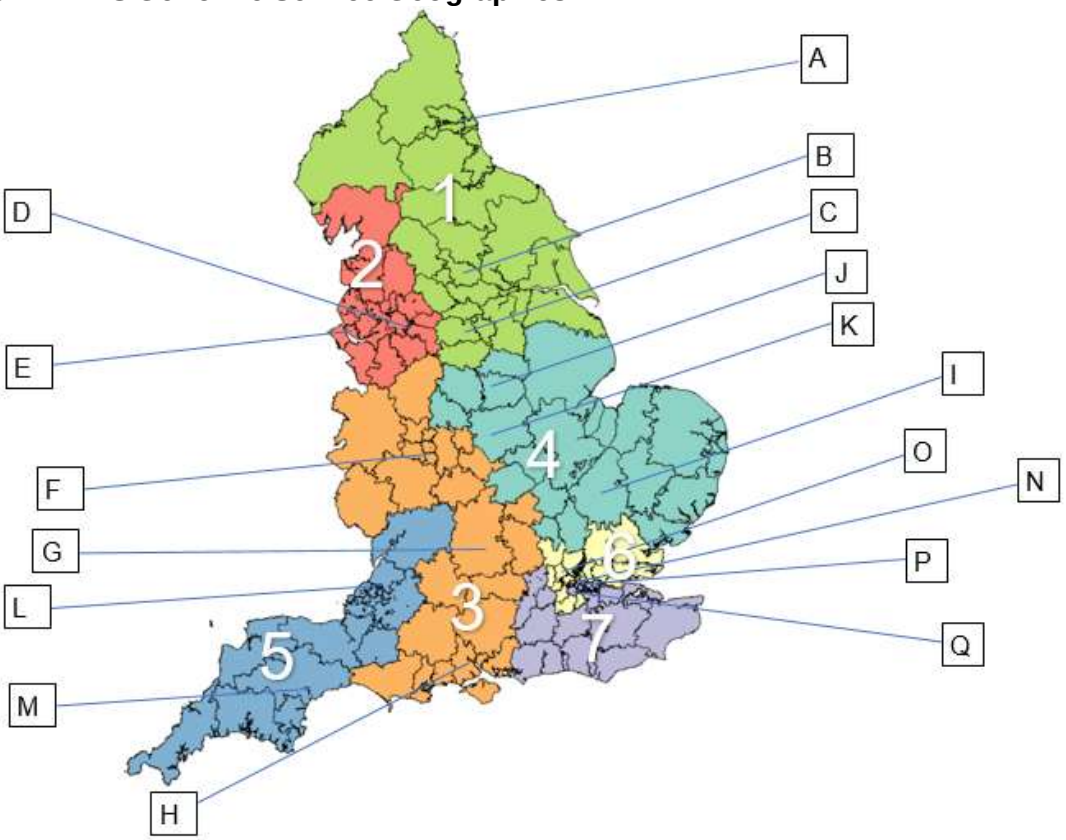
Please refer to the Genomics Clinical Reference Group webpage for NHS England Commissioning Policies which define access to a service for a particular group of service users. The specific clinical policies that relate to the services covered by this service specification include:

- Genetic Testing for BRCA1 and BRCA2 mutations

- Pharmacogenomic testing for DPYD polymorphisms with fluoropyrimidine therapies

Relevant policies/documents:
National Genomic Test Directory
NHS Genomic Medicine Service
NHS Genomic Laboratory Hubs
Rare Disease Collaborative Networks
England Rare Disease Action Plan
Genome UK: the future of healthcare
100,000 Genomes Project
Deciphering Development Disorders
Genomics England's 'Diverse Data' initiative
Joint Committee on Genetics in Medicine - Investing in excellence to provide essential core expertise to the NHS Genomic Medicine Services: Role of the Clinical Geneticist
Joint Committee on Genetics in Medicine – Rationale for job planning document for Consultants in Clinical/Medical Genetics
The Association of Genetic Nurses and Counsellors Guidance Documents
The Association of Genetic Nurses and Counsellors Career Structure for Genetic Counsellors and Support Roles
Genetic Counsellor Registration Board: The Role of the Genetic Counsellor
Referral to Treatment Waiting Time Rules
CQC Regulated Activity: Diagnostic and Screening Procedures
Cancer Registration Statistics, England: 2017
National Genomic Research Library
CanGene CanVar: Data resources, clinical education tools to leverage cancer susceptibility genetics for early detection and prevention of cancer
Best Research for Best Health: The Next Chapter (nihr.ac.uk)
Life Sciences Vision (publishing.service.gov.uk)
Saving and Improving Lives: The Future of UK Clinical Research Delivery (publishing.service.gov.uk)

Annex A. NHS Genomic Service Geographies



Geography Key	Genomic Medicine Service Alliance	Genomic Laboratory Hub	Clinical Genomic Services Key	National Clinical Genomic Service comprised of the individual Regional Clinical Genomics Services
1	North East and Yorkshire	North East and Yorkshire	A	Northern Genetics Service
			B	Yorkshire Regional Genetics Service
			C	Sheffield Clinical Genetics Service
2	North West	North West	D	Manchester Centre for Genomic Medicine
			E	Liverpool Centre for Genomic Medicine
3	Central and South	Central and South	F	West Midlands Regional Genetics Service
			G	Oxford Centre for Genomic Medicine
			H	Wessex Clinical Genetics Service
4	East	East	I	East Anglian Medical Genetics Service
			J	Nottingham Clinical Genetics Service
			K	Leicestershire Clinical Genetics Service
5	South West	South West	L	Bristol Clinical Genetics Service
			M	Peninsula Clinical Genetics Service
6	North Thames	North Thames	N	North East Thames Regional Genetics Service
			O	North West Thames Regional Genetics Service
7	South East	South East	P	South East Thames Regional Genetics Service
			Q	South West Thames Regional Genetics Service