Specialised Services
Service Specification: CP99

Genomics Service

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## Document information

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Statement

Welsh Health Specialised Services Committee (WHSSC) commission clinical and laboratory genomic services in accordance with the criteria outlined in this document.

The All Wales Medical Genomics Service (AWMGS) delivers all diagnostic genomic testing for the Welsh population.

In creating this document WHSSC has reviewed the requirements and standards of care that are expected to deliver this service.

Disclaimer

WHSSC assumes that healthcare professionals will use their clinical judgment, knowledge and expertise when deciding whether it is appropriate to apply this document.

This document may not be clinically appropriate for use in all situations and does not override the responsibility of healthcare professionals to make decisions appropriate to the circumstances of the individual patient, in consultation with the patient and/or their carer or guardian.

WHSSC disclaims any responsibility for damages arising out of the use or non-use of this document.
1. Introduction

This document has been developed as the Service Specification for the planning and delivery of clinical and laboratory genomic services for people resident in Wales. This service will only be commissioned by the Welsh Specialised Services Committee (WHSSC) and applies to residents of all seven Health Boards in Wales. The All Wales Medical Genomics Service delivers all diagnostic genomic testing for the Welsh population.

The All Wales Medical Genomics Service (AWMGS) delivers all diagnostic genomic testing for the Welsh population.

The word genomics within this document refers to genetics and genomics.

1.1 Background

Medical genomics services (both clinical and laboratory) provide patient centred, specialised services focused on the provision of diagnosis and advice to promote improved clinical management and quality of life for those affected by or at risk of a genetic disorder following value based healthcare principles.

Individuals and families are helped to understand their condition, its implications, and their options with regard to reproduction, screening, prevention and management.

Laboratory genomics services are for both rare disease and cancer disorders.

It is anticipated that around 150,000 people in Wales have a rare disease. These are conditions where a pathogenic variant(s)/rearrangement of the DNA (deoxyribonucleic acid) is sufficient to cause the disease. These include haematological disorders such as haemoglobinopathies, sudden cardiac death and inherited metabolic disorders. There are also children with rare diseases where the diagnosis is unclear and where appropriate genomic screening and data collection would significantly improve health and wellbeing outcomes.

Acquired disorders are disorders where an individual has acquired pathogenic variants in a gene or group of genes during their life. Such variants are not inherited from a parent, but occur either randomly or due to environmental exposure. These include most cancers, including haematological cancers.

Genomic services enable clinicians and patients to confirm diagnoses, predict prognosis, and increasingly to determine the most appropriate treatment strategies (known as personalised or stratified medicine).

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1 RDIG

Welsh Health Specialised Services Committee (WHSSC) June 2022
Genomic services fall across three main areas:

- Clinical Genetics
- Laboratory Genomics
- Pathology molecular diagnostic services; these services are not designated as specialist services and are therefore not included in this specification.

1.1.1 Clinical Genetics

Clinical Genetics\(^2\) is the medical specialty which provides a diagnostic service and "genetic counselling" for individuals or families with, or at risk of, conditions which may have a genetic basis.

Genetic counselling is the process through which knowledge about the genetic aspects of illnesses is shared by trained professionals with those who are at an increased risk of either having a heritable disorder or of passing it on to their offspring. A genetic counsellor provides information on the inheritance pattern of illnesses and their recurrence risks, addresses the concerns of patients, their families, and their health care providers, and supports patients and their families dealing with these illnesses\(^3\).

Genomic studies are identifying disease susceptibility variants, that when combined, can generate a polygenic risk score, which indicates the genetic contribution to a common condition, such as breast cancer. The polygenic risk scores will be used in clinical practice to determine disease aetiology, define risks to relatives and select optimum clinical management.

Genetic disorders include:

- Chromosomal abnormalities, which cause birth defects, intellectual disability and/or reproductive problems.
- Single gene disorders such as cystic fibrosis, muscular dystrophy, Huntington's disease and sickle cell disease.
- Familial cancer and cancer-prone syndromes such as inherited breast or colorectal cancer and neurofibromatosis.
- Birth defects with a genetic component such as neural tube defects and cleft lip and palate.

In clinical genetics, the fundamental unit of responsibility is the “family” and includes not only the affected individual who presents for diagnosis and treatment, but also relatives who are identified as being at risk. For example, whilst an individual who presents with ill health needs to be diagnosed and treated within a traditional NHS model, the awareness of family and the relationships within it ensures that the ‘at risk but well’

\(^2\) Clinical Genetics Society
\(^3\) WHO | Genetic counselling services
relatives can be managed appropriately, offering the opportunity for predictive and carrier testing, screening, early intervention and prenatal/preconception genetic counselling.

This specification covers the provision of a National Clinical Genomics Service in Wales.

A Clinical Genetics Service provides:
- clinical assessment and genomic counselling for referred patients
- clinical input to the Clinical Genomics and other clinical specialty MDTs (multi-disciplinary team) as appropriate
- implementation of genomics in clinical pathways throughout other specialties i.e. mainstreaming, in collaboration with colleagues from those specialties
- lifelong management and treatment of patients with rare genomic diseases, in partnership with colleagues in other clinical specialities
- collaboration with colleagues in oncology where needed to support implementation and interpretation of tumour specific genomic testing
- workforce development, including support of education and training of both genetics and non-genetics health professionals across Wales
- participation in clinical genomics related research studies and clinical trials.

1.1.2 Laboratory Genomics

A genetic/genomic test can confirm if there is a clinical pathogenic variant in a particular gene or chromosome, and is carried out on various tissue sample types. When a patient sample is sent to the laboratory, various techniques are used to analyse the sample received.

Laboratory genomics services provide genomic testing services for both rare disease (germline) and cancer. These are provided by the All Wales Genomic Laboratory (AWGL) service which is part of the All Wales Medical Genomics Service (AWMGS).

Services provide gene testing up to the level of large gene panels/whole exome sequencing in addition to whole genome sequencing.

Rare diseases

Services include:
- prenatal testing for at-risk pregnancies and abnormal fetal ultrasound
- postnatal testing for conditions such as developmental delay and infertility
- specific testing for a variety of syndromes and single gene disorders
• predictive testing where a familial pathogenic variant mutation is known, carrier testing, gene screening
• and post-mortem testing for miscarriages.

Cancer
Diagnostic and prognostic testing for:
• haematological malignancy (including post-transplant chimerism monitoring)
• solid tumours
• familial cancer testing
• precision medicine (prognostic stratification, monitoring of treatment response and surveillance).

The laboratory service works closely with the clinical genetic counselling service so that people receive suitable advice both before and after testing.

1.1.3 All Wales Medical Genomics Service (AWMGS)
All Wales Medical Genomics Service (AWMGS) is an NHS specialist regional health service to help and support people in Wales who have genetic conditions, who may be at risk of developing a genetic condition or who are concerned about their family history. They offer advice, information and support to people living in Wales who have questions about their health or their family history. Helping them to understand the possibility of there being a genetic disorder in their family and what this means or could mean for them and their relatives.

The AWMGS laboratory provides a comprehensive state of the art, high throughput, high quality and standardised genetic testing service. They work closely with other specialist teams across mainstream hospital services, including, prenatal, neuromuscular, metabolic disease, endocrine, cardiology and cancer services, and liaise with primary and secondary care services. AWMGS are also involved in the education of other healthcare staff across Wales.

1.2 Aims and Objectives
The aim of this service specification is to define the requirements and standard of care essential for delivering clinical and laboratory genomic services.

The objectives of this service specification are to:
• detail the specifications required to deliver genomic services for people who are resident in Wales
• ensure minimum standards of care are set for the use of genomic services
• ensure value based and equitable access to genomic services
• identify centres that are able to provide genomic services for Welsh patients
• improve outcomes for people with cancer and rare and inherited diseases by accessing genomic services.

1.3 Relationship with other documents
This document should be read in conjunction with the following documents:

• NHS Wales
  o All Wales Policy: Making Decisions in Individual Patient Funding requests (IPFR).
  o Referral Guidelines for Individuals with a Family History of Cancer
  o Referral Guidelines for Individuals with a Personal History of Cancer

• WHSSC policies and service specifications
  o PP184 Genomic Testing Policy Position, June 2020
  o CP37 Pre-implantation Genetic Diagnosis (PGD), August 2014

• National Institute of Health and Care Excellence (NICE) guidance
  o Familial breast cancer: classification, care and managing breast cancer and related risks in people with a family history of breast cancer CG164, Published June 2013, Updated November 2019
  o Molecular testing strategies for Lynch syndrome in people with colorectal cancer DG27, February 2017

• Relevant NHS England policies
  o Medical Genetics Service Specification 2013

• Other published documents
  o Welsh Government, Genomics for Precision Medicine Strategy, July 2017
  o Welsh Rare Diseases Implementation Plan 2017
  o Association for Clinical Genomic Science, General Genetic Laboratory Reporting Recommendations, v1.1 2020
  o Genome UK: The Future of Healthcare 2020
2. Service Delivery

The Welsh Health Specialised Services Committee commission clinical and laboratory genomic services for the people of Wales in line with the criteria identified in this specification.

2.1 Access Criteria

Referrals will be accepted for individuals with a personal or family history of a known genetic disorder, and those individuals with a suspected genetic disorder for whom a clinical genetics assessment is appropriate. Referrals will also be accepted for women in pregnancy where the fetus is affected by, or at risk of, a genetic disorder. In some cases there may be specific referral criteria e.g. cancer genetics. Please refer to the AWMGS website.

Referrals for genetic testing will be accepted for patients that meet the testing criteria for the NHS England National Test Directories and local NHS Wales protocols.

To support the National Genomic Test Directory for Rare and Inherited Disease, testing criteria have been developed and can be found on the NHS England website. The eligibility criteria document supplements the National Genomic Test Directory by setting out which patients should be considered for testing under that indication, and the requesting specialties is a list of the clinical specialties who are permitted to request the test.

The National Test Directory for Cancer includes the eligibility criteria and can be found on the NHS England website.

2.2 Service description

In addition to the standards required within the Contract, specific quality standards and measures will be expected. The AWMGS will monitor compliance of itself against both the professional and local Health Board Informatics and Governance guidance, for example, appropriate data sharing and data privacy. The provider must also meet the standards as set out below.

Medical Genomics services cover both adults and children and will comprise both Clinical Genetic services and Laboratory Genomics services. Medical Genomics services will:

- provide expertise and information for primary and secondary care staff and other health professionals, including the interpretation of laboratory reports conveying complex genetic/genomic results and data, both pathogenic variants and those of uncertain significance
- participate in local and national clinical networks to inform best practice e.g. MDT meetings, Wales cancer network, national

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4 WHSSC Policy Position Statement PP184 for Genomic Testing
dysmorphology meetings, Huntington’s Disease (HD) consortium, Antenatal Screening Wales, the British Society for Genetic Medicine (BSGM) and its constituent groups

- provide a service that meets the needs of patients and their families, as monitored through validated patient satisfaction surveys
- provide formal training programmes and ongoing professional development for clinical geneticists, genetic counsellors and healthcare scientists/practitioners
- provide training and education for other healthcare professionals
- provide training and support for internal continuing professional development and revalidation of staff
- participate in service review meetings, to include laboratory staff, clinical staff and support staff
- participate in External Quality Assessment schemes
- ensure compliance with all national statutory regulatory and external quality accreditation standards
- contribute to Welsh and UK genetic policy through any mechanisms as agreed by WHSSC to provide general advice to WG and WHSSC
- support specialist clinical digital systems and ensure integration with wider NHS Wales’ digital infrastructure; the service will also support the use of modern computing equipment, digital architecture and systems that run on a manufacturer supported version of all relevant software to ensure a secure, safe and efficient operation of genomic testing
- support bioinformatic validation and interpretation for all types of variant (the sensitivity of the variant calling process to be within acceptable national, and where relevant, international standards for the variant type and clinical indication)
- support high throughput, technological and analytical platforms that demonstrate quality outputs and optimised and efficient processing, for example automated nucleic acid extraction etc
- evaluate, procure and validate innovative novel and emergent technologies which deliver best value based healthcare outcomes
- support national Research, Development and Innovation programme
- lead the development of new diagnostic methodologies and technologies for the service, and take part in any future or any potential national genomic projects and initiatives.

**Clinical Genetic Services**

Clinical Genetic Services will:

- provide diagnosis and advice about management of inherited disease and genetic disorders affecting all ages
- provide relatives with risk information and advice about possible interventions to reduce their risk of a poor clinical outcome
• communicate information about the natural history, complications and, where appropriate, management of inherited disease to the patient, relatives and relevant professionals by a variety of means (e.g. letters, telephone, information leaflets)
• provide genetic counselling and support to address the psychosocial aspects of inherited conditions
• offer predictive genetic testing of at-risk relatives for conditions where a familial pathogenic variant has been identified, using agreed protocols where available
• explain the reproductive options available, when appropriate, to those who might wish to receive an early diagnosis or reduce their risk of having an affected child
• offer follow-up, support and advice about health surveillance/screening for specific genetic conditions
• maintain family genetic records, with appropriate information governance protocols
• liaise with the Laboratory Genomics service
• participate in local and national genetic networks
• liaise with other health professionals in order to ensure that the potential benefits and limitations of genetic/genomic testing are understood and that appropriate testing is made available to patients
• participate in MDT meetings with non-genetics clinicians including:
  o fetal medicine MDT
  o psychiatric genetics MDT
  o renal MDTs
  o cardiac MDTs
  o neurosurgery and urology MDTs (VHL)
  o paediatric oncology.
  o various cancer site MDTs.

Clinical Genetic services will be predominantly outpatient based. Initial appointments with a Genetic Counsellor and/or Clinical Genetics Doctor may be held face-to-face, where considered appropriate, or virtually via telephone or an online platform (e.g. Attend Anywhere).

The service will offer an on-call service for urgent advice (e.g. discussion about an abnormal prenatal result), which is available to clinicians and patients across Wales. It will also offer ward consultations (including neonatal and intensive care units) and rapid access clinics for urgent cases.

Wherever possible face to face appointments will take place in the hospital closest to the patient’s home although travel to the host Health Board may be required in some cases.
In addition to general (including cardiac), cancer and prenatal genetics clinics the service should hold specialist clinics covering:

- Rett syndrome
- Tuberous Sclerosis
- Huntington’s disease
- Neuromuscular disorders.

Joint clinics should also be held with other specialities including:

- breast and reconstructive surgery (hereditary breast cancer)
- immunology (22q11 deletion syndrome)
- paediatric oncology
- paediatric cardiology
- neurosurgery (NF2)
- psychiatry (psychiatric genetics).

Where appropriate background information will be gathered by Family History Co-ordinators prior to a patients definitive consultation. This may include:

- detailed family history
- confirmation of diagnoses (e.g. from Cancer Registry)
- review of medical records
- psychosocial circumstances
- preliminary genetic test results.

A proportion of the referrals will be managed by Genetic Counsellors under varying levels of supervision depending upon competence, experience and professional registration as part of a competence framework and scheme of delegation.

Genetics appointments should be of sufficient length to ensure adequate time to provide information and counselling and to enable the patient/relatives to discuss issues fully. These appointments may be face-to-face in the clinic setting, held virtually via Attend Anywhere or by telephone.

The service’s clinicians will access tests for the conditions prescribed in the NHS England test directory as well as NHS Wales agreed protocols.\(^5\)

Laboratory Genomics Services

Laboratory Genomics Services will:

- provide genomic testing, clinical interpretation, reporting, and returning of results within relevant turnaround times for:
  - confirmation or exclusion of diagnosis, this will include provision to support MDT where specialist input is required
  - predictive and confirmatory testing in at-risk family members
  - prenatal testing
  - prenatal screening
  - carrier testing
  - precision medicine for diagnostic and prognostic testing, including for prognostic stratification, monitoring of treatment response and surveillance
  - support for clinical trials and research projects
  - long term banking of DNA (under the terms of the Human Tissue Act)
- contribute clinically significant genetic variant information to appropriate data repositories
- carry out research and development for:
  - introduction of new technologies
  - validation and introduction of new genomic and precision medicine services;
  - development and delivery of translational research projects.
- participate in local and national genetic networks
- determine and implement the most clinically and cost effective means of delivering genomics services (e.g. targeted analysis/gene panels/sequential tests)
- plan services for integrating molecular testing in pathology and clinical genetics into mainstream specialities
- liaise with colleagues: Clinical Scientists support colleagues across the medical specialties in order to ensure that the potential benefits and limitations of genomic testing are understood and made available to patients in all areas of medicine (referred to as ‘mainstreaming’).
- demonstrate there is a sufficiently skilled workforce to deliver, interpret and report timely genomic tests.

Governance and organisational structure

The Medical Genomics service will have an agreed governance and operational framework, which includes:

- Managing Director
- Clinical Director
  - Lead Genetic Counsellor
• Laboratory Director
  o Lead for Cancer
  o Lead for Rare disease
  o Bioinformatics Lead
• Directorate Manager

**Specialist Teams**

• Clinical Geneticists with a specified lead
• Genetics Counsellors with a specified lead
• Genetic Technologists
• Clinical Scientists
• Bioinformaticians.

The service will work with the relevant professional bodies (such as British Society for Genetic Medicine) to create, maintain and provide workforce development plans that demonstrate how the service will provide ongoing professional development for its workforce, and ensure the workforce meets the needs of the Welsh population.

The service will ensure that all staff have up to date professional regulation with the appropriate regulatory body (statutory and non-statutory) and are undertaking professional development and regular revalidation or competency assessment. Career structures and pathways will be aligned with the relevant professional body. Career progression is recognised as vital to the delivery of optimal clinical services.

2.3 **Interdependencies with other services or providers**

• Pathology
• Oncology
• Paediatric
• Clinical psychology
• Screening services (Breast Test Wales, Bowel Screening Wales, Antenatal Screening Wales)
• NHS England/Scotland/Northern Ireland
• Wales Gene Park
• Health Education and Improvement Wales
• All Wales Genomics Oncology Group (AWGOG)\(^6\)

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2.3.1 Relationship between AWGL and health board pathology departments

Health board cellular pathology laboratories receive referrals for genomic testing. There, they will retrieve the tissue block, clean the microtome, cut and mount the sections and the consultant cellular pathologist will mark the tumour on the slides before sending to AWGL for genomic testing\(^7\). Annex ii provides a pictogram of the genomic diagnostic pathway.

The genomic service will engage with health board pathology departments when planning new genomic services.

2.4 Exclusion Criteria

The following exclusions apply to the clinical genetics service:

- People with genetic disorders which have been commissioned as specialised services.

- Individuals with a low likelihood of a monogenic or chromosomal explanation for their health problems e.g. hypermobility without other features.

- Individuals with genetic disorders routinely managed in other clinical specialities e.g. haemophilia.

- Carriers of inherited disorders which fall outside of the scope of the National Genomic Test Directory.

- Carriers of variants predisposing to autosomal recessive diseases detected as an additional finding during the course of investigation, including haemochromatosis and alpha-1 antitrypsin deficiency. Partners should be offered testing according to eligibility in the National Genomic Test Directory.

- Patients requiring primary investigation of recurrent miscarriage and male infertility.

- Individuals with acquired (somatic) variants identified in tumours that are known to be absent from the germline; if the variant is definitely or potentially present in the germline it may be appropriate to seek advice or make a referral to clinical genetics.

- Individuals with results from commercial direct to consumer genomic tests which do not meet the referral criteria\(^8,9\).

\(^7\) This element of the pathway is not commissioned by WHSSC but is the commissioning responsibility of health boards.

\(^8\) Please also see the All Wales Prior Approval Policy section on “Transfer back to the NHS following self-funding in the private sector”

\(^9\) The Royal College of General Practitioners (RCPG) and The British Society for Genetic Medicine (BSGM) position statement on this matter can be found at: https://www.rcgp.org.uk/-/media/Files/CIRC/Clinical-Policy/Position-statements/RCPG-position-statement-on-direct-to-consumer-genomic-testing-oct-2019.ashx?la=en
• Referrals for genetic testing from private providers to the Clinical Genomics Services which fall outside of the defined criteria within this document\(^6\).

• Provision of advice on the results of private genomic tests performed on tumours\(^6,10\)

• Provision of advice on privately procured genomic test results where variants are reported as pathogenic outside of the NHS and inappropriate action advised to patients\(^6\).

### 2.5 Acceptance Criteria

The service outlined in this specification is for patients ordinarily resident in Wales, or otherwise the commissioning responsibility of the NHS in Wales. This excludes patients who whilst resident in Wales, are registered with a GP practice in England, but includes patients resident in England who are registered with a GP Practice in Wales.

### 2.6 Patient Pathway

Given the diversity of the diseases seen by medical genomics, there is no generic pathway for access into/ through the services.

### 2.7 Referrals to the Clinical Genetics Service

Referrals may originate from all areas of medicine, most commonly general practice, paediatrics, surgery and oncology. Patients can access clinical genetics via referrals from the following areas:

- Primary Care i.e. General Practitioners (GPs) or other healthcare professionals
- Secondary Care i.e. Consultants/Services/appropriate clinicians
- Tertiary Care i.e. Specialist Services
- Self/Family i.e. relatives of existing AWMGS patients

Self-referrals are received from individuals and families who are already known to the service, contact is usually made when family circumstances change e.g. pregnancy or a new diagnosis of a cancer. The referral pathway into the service will depend upon the nature of the condition found in the patient’s family.

Additional family members for whom the discussion is of potential relevance may also attend appointments on an exceptional basis, self-referrals will be accepted, if there is clear evidence of a familial condition and a GP has been approached but declined to refer. Any such cases will be audited to ensure these criteria have been met – and are not expected to exceed 1% of referrals.

\(^{10}\) Note that there may be some instances where the provision of advice is appropriate, e.g. where a specific genetic test is required as a condition of entering a clinical trial
Referrals may be urgent, including PICU/NICU ward referrals, prenatal referrals and oncology referrals.

The Clinical Genetics service will have a process in place to ensure patients for whom a genetics assessment is likely to be beneficial are accepted.

The Clinical Genetic services will have a process in place to determine how referrals will be managed. These will describe who is the most appropriate person to manage a case (genetic counsellor or clinical geneticist) and will explain which patients need to be offered a clinic appointment. The process will also determine the modality of the clinical contact (face-to-face/telephone/video clinic), while ensuring the appropriate flexibility to meet local needs.

2.8 Referrals to the Genomics Laboratory service

The Genomics Laboratory service will create, implement and maintain genomic testing referral pathways that cover each health board in Wales.

The Laboratory Director will be responsible for determining the approval process and that the request falls inside the scope of the commissioning model. It is the responsibility of the Laboratory Director to manage any test request that falls outside of the commissioning model.

Clinical services will refer patients for tests which they believe will contribute to the diagnosis or management of the patient’s condition. Referrals for patients and families with inherited disorders should take account of the National Genomic Test Directory or agreed local NHS Wales eligibility criteria, where these are available. For acquired disorders, the appropriate clinical referral guidelines should be followed. Where the National Genomic Test Directory or agreed local NHS Wales eligibility criteria are available these should be adhered to in the ordering of tests.

Where the service cannot provide a test, the laboratory will follow the send-out procedure for transporting specimens to other laboratories, receiving results back safely and relaying these to the requesting clinician. The service will routinely repatriate send-away tests where it is economically viable to do so.

See also:
- Rapid Whole Genome Sequencing referral guidelines.
- Welsh Health Specialised Services Policy Position Statement PP184 for Genomic Testing
2.9 Service provider/Designated Centre

- All Wales Medical Genomics Service
  University Hospital of Wales
  Heath Park
  Cardiff
  CF14 4XW

2.10 Exceptions

If the patient does not meet the criteria for treatment as outlined in this policy, an Individual Patient Funding Request (IPFR) can be submitted for consideration in line with the All Wales Policy: Making Decisions on Individual Patient Funding Requests. The request will then be considered by the All Wales IPFR Panel.

If the patient wishes to be referred to a provider outside of the agreed pathway, an IPFR should be submitted.

Further information on making IPFR requests can be found at: Welsh Health Specialised Services Committee (WHSSC) | Individual Patient Funding Requests
3. Quality and Patient Safety

The provider must work to written quality standard and provide monitoring information to the lead commissioner. The quality management systems must be externally audited and accredited.

The centre must enable the patients, carers and advocates informed participation and be able to demonstrate this. Provision should be made for patients with communication difficulties and for children, teenagers and young adults.

3.1 Quality Indicators (Standards)

The aim is to provide a comprehensive, co-ordinated and resilient medical genomics service.

The quality indicators will be aligned with Welsh Government health and social care strategy, A Healthier Wales: Long Term Plan for Health and Social Care.

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<th>Indicator</th>
<th>Extended Description</th>
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<td>A patient experience exercise is undertaken at least every two years.</td>
<td>The service has undertaken a patient experience exercise during the last 24 months to obtain feedback on patients and parents/carers experience of the services offered. Evidence to ensure that the patient experience exercise is representative of all sections of the population served. The exercise and any remedial actions taken as a result, should be presented to, and discussed at, the relevant governance group and with outcomes shared with patients and families.</td>
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<tr>
<td>There is information available for patients and/or their parents and carers.</td>
<td>The service makes available/signposts written (leaflet, app or website based) information for patients and parents / carers covering at least the following: - the service offered - information about conditions - support groups, where relevant - signposting to relevant helplines/services/voluntary sector resources where relevant - contact details for the regional CGS and the key personnel. The information is made available in formats appropriate for the population needs - e.g. different languages, Braille etc</td>
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<td>The service has a patient involvement group</td>
<td>The service has a patient involvement group that meets at least annually to provide feedback on</td>
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11 Use the genomic counselling outcome scale (Grant et al, Eur J Med Genet. 2019;62:324-334)
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<td>services and new developments. The constitution of the group should reflect the regional population in terms of age, ethnicity, geography, gender. Evidence that the views of the group are acted upon and incorporated into service plans.</td>
<td>Self-referral to the clinical genomic service is facilitated. In advance of national guidance for accepting and rejecting self-referrals being established, the provider will ensure the provision of an equitable service.</td>
</tr>
<tr>
<td>Self-referral to the clinical genetic service is facilitated</td>
<td>Different methods of patient interaction are offered. The service will offer at least face to face, telephone and video clinic appointments to ensure improved access to service.</td>
</tr>
<tr>
<td>Different methods of patient interaction are offered</td>
<td>The clinical genomic service has team members as per the service specification.</td>
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<tr>
<td>The service has team members as per the service specification.</td>
<td>The lead Clinical Geneticist and lead Genetic Counsellor will provide leadership and oversight for the service. They will represent their centre at national meetings and work in a networked approach with other services to ensure equity and excellence in provision of services.</td>
</tr>
<tr>
<td>The service has a lead Clinical Geneticist and lead Genomic Counsellor</td>
<td>The service has an operational plan which details the categories of personnel required to support genomics in MDTs in rare diseases and inherited cancers.</td>
</tr>
<tr>
<td>The service has an operational plan for genomic led MDTs</td>
<td>There is a clinical genomic multi-disciplinary team meeting held on a minimum monthly basis to discuss complex clinical diagnosis/management for patients. A SOP is in place and adhered to for each MDT The meeting includes at least the following MDT members, (the quorum). - Clinical Geneticists and; - Genetic Counsellors Other professionals including Clinical scientists, Trainees, Allied Health Professionals and nurses should be encouraged to attend The clinical management decisions taken and the meeting attendance is recorded in the patient record. There is a policy where urgent cases are considered without waiting for the next MDT if clinical need dictates. Referrals to the MDT will be as outlined within the service specification.</td>
</tr>
<tr>
<td>A multi-disciplinary team meeting is held at a minimum of a monthly basis.</td>
<td></td>
</tr>
<tr>
<td>Indicator</td>
<td>Extended Description</td>
</tr>
<tr>
<td>--------------------------------------------------------------------------</td>
<td>----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------</td>
</tr>
<tr>
<td>There are multi-disciplinary clinics for patients with complex needs and to support local management of patients with less complex needs.</td>
<td>The provider will offer multidisciplinary clinics to ensure patients with complex needs are referred to clinical genetics and support local management of those with less complex needs, and/or availability of expert advice by phone, video link or e-mail.</td>
</tr>
<tr>
<td>The service will ensure that they are providing services commensurate with the training and skills of their staff</td>
<td>The provider will deliver GC led clinics in many sub-specialty areas of genomics including cancer and cardiac genetics as part of an overall service, supported by the whole team. Each service will demonstrate evidence of career progression for staff within the service.</td>
</tr>
<tr>
<td>The service will undertake a survey at least every two years to determine the value of the service to primary, secondary and tertiary health care providers</td>
<td>A survey is held every two years to capture the views of health professionals, - doctors, nurses and AHPs in other specialties and primary care to determine the impact of the service in supporting mainstreaming and providing expert support to facilitate local care</td>
</tr>
<tr>
<td>The service has a process in place for identifying specific training needs in genomics</td>
<td>The service will work with Health Education and Improvement Wales, Royal College of Physicians, GMS Alliance, the professional bodies (British Society of Genomic Medicine, Clinical Genetics Society and Association of Genetic Nurses and Counsellors) to create, maintain and provide workforce development plans that demonstrate how the service will provide ongoing professional development for its workforce. Due to development of the specialty a large number of consultant clinical geneticists are due to retire in the next decade. The service will work with other stakeholders to identify skill and knowledge gaps within both the service and broader health workforce and will support its partners to create a sufficiently skilled workforce to deliver the service, will support the education and training to new and existing healthcare science workforce and the broader workforce to make the best use for the NHS of the expertise in genomic technologies and its application for patient benefit. This will include education in primary and secondary care to enable those services to identify moderate risk familial cancer patients to facilitate appropriate screening, and referral for those patients at higher familial risk who require expert input.</td>
</tr>
<tr>
<td>The service will have a policy to provide urgent clinical input for prenatal and neonatal services</td>
<td>The service should ensure that provision is in place to initiate contact and clinical input to a referred individual within two working days for an urgent referral - i.e. where support is required to inform genetic testing or advice during pregnancy or to guide genomic testing in an acutely unwell neonate on NICU</td>
</tr>
<tr>
<td>Indicator</td>
<td>Extended Description</td>
</tr>
<tr>
<td>-----------</td>
<td>----------------------</td>
</tr>
<tr>
<td>There are agreed patient pathways as per the service specification.</td>
<td>Work with other clinical specialties to ensure that the provider has patient pathways for referral, transfer, treatment, shared care, and follow up. These will include: - Referral pathways to the NCGS for: - diagnosis, - screening, - investigation, - management, - risk assessment, including predictive testing - genomic counselling and psychosocial support of patients with complex, rare and undiagnosed conditions</td>
</tr>
<tr>
<td>There are transition pathways in place.</td>
<td>There is a policy to review notes/clinical cases where transition arrangements/pathways for young people are required, i.e. where decisions regarding reproductive choices, ongoing screening, consideration of genetic testing are relevant.</td>
</tr>
<tr>
<td>There are agreed clinical guidelines as per the service specification.</td>
<td>There is adherence to agreed clinical guidelines (including NICE Guidelines e.g. for breast cancer, Lynch syndrome, familial hypercholesterolaemia) which reflect the service specification and national and international best practice. That guidance from Professional bodies (BSGM/CGS/AGNC etc) and Royal Colleges is adopted</td>
</tr>
<tr>
<td>The provider participates in local and national audits.</td>
<td>The provider participates in local and national (including Professional Society) audits as required. Audits of quality of letters summarising patient consultations are undertaken - to record following information diagnosis (if known) is recorded, reason for consultation, referrer details, outcome - next steps, need for genetic testing, time frame for results if appropriate. The service will undertake an audit of compliance with recording consent/record of discussion for individuals undergoing genetic testing. The outcomes of the audit are fed back to any relevant governance meetings as required.</td>
</tr>
<tr>
<td>The provider reviews annually its contribution to research and clinical trials.</td>
<td>The provider reviews annually the availability and participation in clinical trials (interventional and observational) including: - for each clinical trial and well-designed study the number of patients participating (CRN listed studies) or the reason why recruitment has not been possible - future plans to improve recruitment.</td>
</tr>
<tr>
<td>The service has an operational policy for storage and usage of genomic data</td>
<td>The service will have an operational policy for storage and usage of genomic data</td>
</tr>
<tr>
<td>Indicator</td>
<td>Extended Description</td>
</tr>
<tr>
<td>--------------------------------------------------------------------------</td>
<td>------------------------------------------------------------------------------------------------------------------------------------------------------</td>
</tr>
<tr>
<td>The service participates in National and Regional meetings relevant to clinical care</td>
<td>Each CGS will participate in National MDT meetings and in National and Regional meetings to discuss patient optimum management.</td>
</tr>
<tr>
<td>The service will have a policy in place to capture health inequalities in access to the service</td>
<td>The service will have a policy in place to capture health inequalities in access to the service.</td>
</tr>
</tbody>
</table>

### 3.2 Other quality requirements

- Patients/families should be provided with accurate, up-to-date information on genetic risks, testing and/or screening, and advised about reproductive choices that are available, with information discussed during face-to-face, virtual or telephone consultations summarised afterwards in writing.
- Clinicians should usually write to patients directly to explain complex terminology/concepts, or copy clinic letters to them and ensure effective patient involvement in all decision making.
- Different methods of patient interactions should be offered, with patients being given a choice where this is appropriate.
- Consent for genetic testing and retention of deoxyribonucleic acid (DNA), cell free DNA and ribonucleic acid (RNA) samples must be undertaken in accordance with Department of Health guidance and the Mental Capacity Act, 2005.
- Education should be provided during clinic, through post-clinic letters and via patient information leaflets (in different languages). Information is available for patients and/or their parents and carers.
- Professional interpreting services, including signing interpreters, should be used if necessary.
- Patients and families should be directed to relevant lay support groups including the Genetic Alliance UK and UNIQUE (Rare Chromosome Disorder Support Group).
- Services should recognise the role of the carer, which is vital for many patients, especially those with learning disability and those with physical or psychiatric impairment. Advocacy and support should be offered to carers appropriately.
- The service will comply with all relevant statutory regulatory standards
- The service should participate in National and Regional meetings relevant to clinical care.
- All adverse incidents should be thoroughly investigated and reported along appropriate lines either internally within Cardiff and Vale UHB or to WHSSC and or Welsh Government if they meet the criteria of a
Serious Adverse Event. Learning from adverse events will be shared either locally or nationally as appropriate

- The service will comply with NHS Wales commissioning policies and policy statements.
- The provider will have a recognised system to demonstrate service quality and standards including audit, training, quality assurance, equipment maintenance, staff competence, incident reporting.
- The service will have detailed clinical protocols setting out nationally (and locally where appropriate) recognised good practice for each clinic site.
- Genomic laboratories will have systems in place for managing test requests, which allows them to be processed in a clinically appropriate, cost effective and timely way.
- The laboratory is responsible for the storage of all samples which should be following current NHS diagnostic laboratory practices and in line with the guidance from “the Royal College of Pathologists and the Institute of Biomedical Science/the retention and storage of pathological records and specimens”.
- The quality system and its treatment protocols will be subject to regular clinical and management audit.
- Genetics laboratories must be accredited to the International Organization for Standardization (2012) Medical laboratories – Requirements for quality and competence (ISO Standard No. 15189:2012) by the United Kingdom Accreditation Service (UKAS). UKAS is the sole national accreditation body for the United Kingdom. UKAS is recognised by government, to assess against internationally agreed standards, organisations that provide testing services. UKAS, will accredit the service to acknowledge they have demonstrated compliance with the standards when undertaking consistent, accurate, and timely testing procedures and the reporting of test results in accordance with nationally agreed target reporting times appropriate for the nature of the sample and referral reason (see 4.3 Laboratory Reporting Time Targets below).
- The genomics laboratory service should participate in the following external quality assessment schemes:
  - Genomics Quality Assessment (GenQA)
  - European Molecular Genetics Quality Network (EMQN).
4. Performance monitoring and Information Requirement

4.1 Performance Monitoring

WHSSC will be responsible for commissioning services in line with this service specification. This will include agreeing appropriate information and procedures to monitor the performance of organisations.

The AWMGS will provide regular information to the commissioners about the use, activity and performance of their service against key outcomes, as defined below.

For the service defined in this specification, the following approaches will be adopted:

**Clinical Genetics**

The information section of the contract provides the basic individual patient data and activity reporting required as part of the basic billing and contract monitoring arrangements (e.g. new/follow up outpatients and types of test/source of referral).

The following proxy outcome measures will be reported to WHSSC annually, on a quarterly basis for the core services:

- numbers of new and follow up patients seen
- reason for referral broken down into cancer, general and prenatal categories (this will aid future service planning)
- numbers of new referrals, clinics held and risk letters produced
- Did Not Attends as defined in the Data Dictionary - proportion of appointments that are not attended
- clinic breakdown by appointment, pre-clinic, ward, and clinic (face-to-face, virtual or telephone)
- patients consulted by a genetic counsellor - number of patients consulted by a genetic counsellor during period and number of appointments provided by a genetic counsellor during period
- current status of waiting lists
- educational sessions provided by clinical genetics to other specialties to support genetics in mainstream medicine - number of educational sessions provided by clinical genetics to other specialties
- patient experience - number of written complaints about the genetics department and number of letters/emails from patients, carers or non-genetics consultants registering thanks to the genetics department
- significant clinical or regulatory adverse incidents
- novel research, service development and innovation programmes
- financial position
- staffing position
- clinical audits - Proportion of nationally approved clinical audits completed and action plans put in place (the number and type of audits need to be agreed).

**Genomics Laboratory**

The following will be reported to WHSSC annually, on a quarterly basis for the core services:
- number of samples received
- number of samples extracted
- number and type of samples sent out to other laboratories for genomic testing
- financial position
- compliance with professional turnaround times for lab
- serious incidents regarding lab tests - number of serious incidents involving laboratory tests
- highlight critical risks
- benchmarking

The following will be reported to WHSSC by both the clinical genetics service and the genomics laboratory:
- all serious incidents will be reported within two days of incident
- serious incidents regarding patient care, outcomes or experience
- serious incidents regarding laboratory tests
- serious incidents involving regulatory compliance (H&S, HTA etc.)
- significant findings of quality accreditation inspections

### 4.2 Key Performance Indicators

The providers will be expected to monitor against the full list of Quality Indicators derived from the service description components described in Section 2.2.

The provider should also monitor the appropriateness of referrals into the service and provide regular feedback to referrers on inappropriate referrals, identifying any trends or potential educational needs.

### 4.3 Laboratory Reporting Time Targets

Accurate test results should be provided in accordance with the agreed reporting times. Reporting times vary depending on the nature and urgency of the referral. These are taken from the Association of Clinical Genomic Science\(^\text{12}\) (ACGS) and reproduced in the table below.

\(^{12}\) [ACGS Genetic Laboratory Reporting Recommendations 2020 v1.1](#)
All reporting times are given in calendar days.

The reporting time targets are maximum and the aim shall be to report results as soon as practicably possible.

All targets should be for 90% within the given reporting time target for any category.

Day 0 is the day the sample is received into the laboratory with all appropriate information and all other required samples received. This can also be the day that a specific investigation is activated if a request is made by a clinician for a test on a stored sample.

The end point of the test is measured when the results are available in an authorised state. This can be electronically stored and not yet sent out by the laboratory.
### 4.4 Genetic Laboratory Reporting Time Targets

<table>
<thead>
<tr>
<th>Clinical Urgency</th>
<th>Category (mapping to test directory)</th>
<th>Sub-categories</th>
<th>Calendar Days</th>
<th>Examples</th>
</tr>
</thead>
<tbody>
<tr>
<td>URGENT</td>
<td>Ultra-Rapid</td>
<td>N/A</td>
<td>3 days</td>
<td>QF-PCR for rapid trisomy detection</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>Urgent haematology oncology FISH/RT-PCR</td>
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<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>PCR-based tests where the result is needed urgently for prenatal diagnosis</td>
</tr>
<tr>
<td>URGENT</td>
<td>Ultra-Rapid</td>
<td>NA</td>
<td>7 days</td>
<td>NIPT</td>
</tr>
<tr>
<td>URGENT</td>
<td>Rapid</td>
<td>Rapid</td>
<td>14 days</td>
<td>Microarray for prenatal / urgent postnatal (e.g. neonatal referrals)</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>Urgent Haematology oncology karyotyping</td>
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<td></td>
<td></td>
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<td></td>
<td>Mutation specific molecular pathology tests</td>
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<td></td>
<td></td>
<td></td>
<td></td>
<td>Southern blot tests where the result is needed urgently for prenatal diagnosis</td>
</tr>
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<td></td>
<td></td>
<td></td>
<td></td>
<td>PCR-based tests for predictive testing and confirmation of neonatal results</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>Urgent panels and exomes for relevant indications</td>
</tr>
<tr>
<td></td>
<td>Complex rapid</td>
<td></td>
<td>21 days</td>
<td>NIPD</td>
</tr>
<tr>
<td>NON-URGENT</td>
<td>Standard</td>
<td>Somatic Cancer</td>
<td>21 days</td>
<td>Standard HO karyotyping (e.g. MDS)</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>NGS panels for HO referrals</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Rare Disease</td>
<td>42 days</td>
<td>Standard single gene and small gene panel (&lt;10 gene) sequencing</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>(6 weeks)</td>
<td>Known familial mutation testing</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>Standard paediatric microarray</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>NGS panels for molecular pathology referrals</td>
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<td></td>
<td></td>
<td></td>
<td></td>
<td>Standard single gene and small gene panel (&lt;10 gene) sequencing</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>Known familial mutation testing</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>Standard STR based analysis</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>Postnatal karyotyping (e.g. fertility or familial microarray follow-up)</td>
</tr>
<tr>
<td>NON-URGENT</td>
<td>Complex Standard</td>
<td>Rare Disease</td>
<td>84 days</td>
<td>Large gene-panels (&gt;10 genes) or WES for standard referral indications</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>(12 weeks)</td>
<td>Part a) 42 days (6 weeks) Expectation for delivery of centralised WGS (from DNA sample receipt to return of vcf and/or filtered variants to GLH)</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>Part b) 42 days (6 weeks) Validation/reporting of centralised WGS results after receipt at GLH</td>
</tr>
</tbody>
</table>
4.5 Date of Review

This document is scheduled for review before 2025 where we will check if any new evidence is available.

If an update is carried out the policy will remain extant until the revised policy is published.
5. Equality Impact and Assessment

The Equality and Health Impact Assessment (EHIA) process has been developed to help promote fair and equal treatment in the delivery of health services. It aims to enable Welsh Health Specialised Services Committee to identify and eliminate detrimental treatment caused by the adverse impact of health service policies upon groups and individuals for reasons of race, gender re-assignment, disability, sex, sexual orientation, age, religion and belief, marriage and civil partnership, pregnancy and maternity and language (Welsh).

This policy has been subjected to an EHIA.

The Assessment demonstrates the policy is robust and there is no potential for discrimination or adverse impact. All opportunities to promote equality have been taken.
6. Putting Things Right

6.1 Raising a Concern

Whilst every effort has been made to ensure that decisions made under this policy are robust and appropriate for the patient group, it is acknowledged that there may be occasions when the patient or their representative are not happy with decisions made or the treatment provided.

The patient or their representative should be guided by the clinician, or the member of NHS staff with whom the concern is raised, to the appropriate arrangements for management of their concern.

If a patient or their representative is unhappy with the care provided during the treatment or the clinical decision to withdraw treatment provided under this policy, the patient and/or their representative should be guided to the LHB for NHS Putting Things Right. For services provided outside NHS Wales the patient or their representative should be guided to the NHS Trust Concerns Procedure, with a copy of the concern being sent to WHSSC.

6.2 Individual Patient Funding Request (IPFR)

If the patient does not meet the criteria for treatment as outlined in this policy, an Individual Patient Funding Request (IPFR) can be submitted for consideration in line with the All Wales Policy: Making Decisions on Individual Patient Funding Requests. The request will then be considered by the All Wales IPFR Panel.

If an IPFR is declined by the Panel, a patient and/or their NHS clinician has the right to request information about how the decision was reached. If the patient and their NHS clinician feel the process has not been followed in accordance with this policy, arrangements can be made for an independent review of the process to be undertaken by the patient’s Local Health Board. The ground for the review, which are detailed in the All Wales Policy: Making Decisions on Individual Patient Funding Requests (IPFR), must be clearly stated.

If the patient wishes to be referred to a provider outside of the agreed pathway, and IPFR should be submitted.

Further information on making IPFR requests can be found at: Welsh Health Specialised Services Committee (WHSSC) | Individual Patient Funding Requests
## Annex i  Abbreviations and Glossary

### Abbreviations

<table>
<thead>
<tr>
<th>Abbreviation</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>AGNC</td>
<td>Association of Genetic Nurses and Counsellors</td>
</tr>
<tr>
<td>AHCS</td>
<td>Academy for Healthcare Sciences</td>
</tr>
<tr>
<td>AWGL</td>
<td>All Wales Genomics Laboratory</td>
</tr>
<tr>
<td>AWMGS</td>
<td>All Wales Medical Genomics Service</td>
</tr>
<tr>
<td>BRCA</td>
<td>Breast Cancer Gene</td>
</tr>
<tr>
<td>BSGM</td>
<td>British Society for Genetic Medicine</td>
</tr>
<tr>
<td>DNA</td>
<td>Deoxyribonucleic Acid</td>
</tr>
<tr>
<td>EMQN</td>
<td>European Molecular Genetics Quality Network</td>
</tr>
<tr>
<td>EQA</td>
<td>External Quality Assurance</td>
</tr>
<tr>
<td>EHIA</td>
<td>Equality and Health Impact Assessment</td>
</tr>
<tr>
<td>GCRB</td>
<td>Genetic Counsellor Registration Board</td>
</tr>
<tr>
<td>GENQA</td>
<td>Genomics Quality Assessment</td>
</tr>
<tr>
<td>HCPC</td>
<td>Health and Care Professions Council</td>
</tr>
<tr>
<td>HSSE</td>
<td>Higher Specialist Scientist Training</td>
</tr>
<tr>
<td>HSSR</td>
<td>Higher Specialist Scientist Equivalence</td>
</tr>
<tr>
<td>HSST</td>
<td>Higher Specialist Scientist Register</td>
</tr>
<tr>
<td>HTA</td>
<td>Human Tissue Authority</td>
</tr>
<tr>
<td>IPFR</td>
<td>Individual Patient Funding Request</td>
</tr>
<tr>
<td>MDT</td>
<td>Multi-Disciplinary Team</td>
</tr>
<tr>
<td>NHS</td>
<td>National Health Service</td>
</tr>
<tr>
<td>PTP/E</td>
<td>Practitioner Training Programme/Equivalence</td>
</tr>
<tr>
<td>RNA</td>
<td>Ribonucleic Acid</td>
</tr>
<tr>
<td>STP/E</td>
<td>Scientist Training Programme/Equivalence</td>
</tr>
<tr>
<td>UHB</td>
<td>University Health Board</td>
</tr>
<tr>
<td>UKAS</td>
<td>United Kingdom Accreditation Service</td>
</tr>
<tr>
<td>UK NEQAS</td>
<td>United Kingdom National External Quality Assessment Service</td>
</tr>
<tr>
<td>WG</td>
<td>Welsh Government</td>
</tr>
<tr>
<td>WHSSC</td>
<td>Welsh Health Specialised Services Committee</td>
</tr>
</tbody>
</table>
Glossary

**Individual Patient Funding Request (IPFR)**
An IPFR is a request to Welsh Health Specialised Services Committee (WHSSC) to fund an intervention, device or treatment for patients that fall outside the range of services and treatments routinely provided across Wales.

**Welsh Health Specialised Services Committee (WHSSC)**
WHSSC is a joint committee of the seven local health boards in Wales. The purpose of WHSSC is to ensure that the population of Wales has fair and equitable access to the full range of Specialised Services and Tertiary Services. WHSSC ensures that specialised services are commissioned from providers that have the appropriate experience and expertise. They ensure that these providers are able to provide a robust, high quality and sustainable services, which are safe for patients and are cost effective for NHS Wales.
Annex ii Genomic Diagnostic Testing Pathway

**AWMGS LABORATORY**
Sample receipt in genetics lab and DNA macrosection for DNA and RNA

**CELL PATH LAB**
Slide cutting and calculation of tumour content

**DNA extraction**

**NGS**

**Curation**

**Bioinformatic Analysis and interpretation**

**FFPE**

Cancer Patient

**Report**