Pwyllgor Gwasanaethau Iechyd Arbenigol Cymru (PGIAC)
Welsh Health Specialised Services Committee (WHSSC)

Specialised Services Service Specification:
CP99
Genetic Services

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Contents

1. AIM

1.1 Introduction

This document has been developed as the service specification for the planning of clinical and laboratory genetic services for patients and families resident in Wales.

The purpose of this document is to:

- detail the specification for clinical genetic services for patients and families who are resident in Wales;
- detail the specification for genetic testing for patients and families who are resident in Wales;
- to provide patient pathways when available.

1.2 Relationship with other Policy and Service Specifications

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

- All Wales Policy: Making Decisions on Individual Patient Funding Requests (IPFR)
- Specialised service specification CP62: Genetic Testing for inherited Cardiac conditions.
- Specialised Service CP57: Genetic Testing for inherited Cardiac conditions.
- Specialised Service CP37: Pre-implantation Genetic diagnosis.
- Wales Implementation plan (2014) for The UK's Rare Diseases Strategy (2013).
- Genetic Testing for Stratified medicine , Task and Finish Group of the Cancer Blood and Plastics Programme team
- The aims of the Familial Hypercholesterolemia Wales service: http://www.fhservice.wales.nhs.uk/home
- Welsh Governments emerging strategy for genomics in Wales.
- WHSSC Integrated Medium Term Plan
- UK Rare Diseases Strategy
- Welsh Governments 2015 Welsh Implementation Plan for Rare diseases.
- Welsh Government Together for Health cancer delivery plan our vision

2. SERVICE

2.1 Service Model

2.1.1 Aim

The overarching aim of Medical Genetic services (both clinical and laboratory) is to provide a 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 condition or congenital abnormality.

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

Medical genetics is concerned with the diagnosis and management of diseases that are more common and that are not classified as rare diseases, in addition to those rare diseases.

In addition to inherited disorders, Laboratory Genetics provides services for acquired disorders (cancer). These 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)

Between 5,000 and 8,000 rare diseases have been identified within the UK. Each disease affects less than 0.1% of the UK's population, but taken together they affect the lives of 3 million people. Of which, at least 80% have an identified genetic origin.

Genetic services fall across three main areas:

- Clinical Genetics;
- Laboratory genetics;
- Biochemical and pathology services, these services are not designated as specialist services and are not referred to hereafter within this service specification.

It is essential medical genetic services are able to provide integrated clinical and laboratory genetic services that are equitable, safe, efficient, appropriate, accessible, acceptable and responsive to the health care needs of the population of Wales and of a demonstrably high quality.

2.1.2 Clinical genetics

Clinical Genetics provides services for any individual or family affected by, or at risk of, a genetic disorder or congenital abnormality.

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' relatives can be managed appropriately, offering the opportunity for predictive and carrier testing, screening, early intervention and prenatal/preconception genetic counselling.

2.1.3 Laboratory Genetics

Laboratory genetics will provide molecular and cytogenetic analyses for an expanding repertoire of diseases, both inherited and acquired, to a variety of clinicians including clinical geneticists, pediatricians and oncologists. Laboratory scientists will provide genetic analyses with full clinical interpretation to aid the clinical management and treatment of patients and their families.

2.2 Models of Delivery

Medical Genetic services will provide the following:

- Diagnosis or exclusion of inherited disease and genetic disorders affecting all ages;
- Enable other clinician disciplines to utilise genetic and genomic services;
- Targeted investigations, using both specialised genetic and conventional non-genetic tests, (provided by biochemical and pathology services) for the purposes of diagnosis, risk assessment and management;
- Diagnosis and early management of inherited diseases to reduce the morbidity and/or mortality for many of these clinical conditions in those affected; and/or provide relatives with risk information and possible interventions to reduce their risk of poor clinical outcome;
- Communication of the natural history, complications and appropriate management of inherited disease, to the patient, relatives and relevant professionals;

- Genetic counselling and attention to psychosocial aspects of inherited conditions;
- Genetic counselling and effective communication of appropriate genetic information in the clinic and by a variety of means (e.g. letters, telephone, information leaflets);
- Predictive genetic testing of at-risk relatives for conditions where a familial mutation or cause has been identified, using agreed protocols where available;
- Explanation of the reproductive options available, when appropriate, to women/couples who might wish to receive an early diagnosis or reduce their risk of having an affected child;
- Where appropriate, follow-up, support and coordination of health surveillance / screening for specific genetic conditions. Identification of genetic risk to the wider family and, where appropriate, the offer of genetic services to extended family members;
- Formal training programmes and ongoing professional development for clinical geneticists, genetic counsellors and healthcare scientists/practitioners;
- Training and education for other healthcare professionals;
- Laboratory investigations, in support of clinical genetics and many other specialist services, including discussion and interpretation of complex results and data of uncertain significance. Laboratories liaise with other genetics laboratories in local and national networks;
- Participation in service review meetings, to include laboratory staff, clinical staff, consultants, genetic counsellors and nurses. These will form part of the WHSSC planning directorate audit programme;
- Participation in local and national clinical networks, e.g. oncology, neurology, foetal medicine, cardiology; national dysmorphology, Cancer Genetics Group meetings, UK Huntington Disease Consortium and Genetics meeting to inform best practice;
- Provision of expertise and information for other secondary and primary care staff and other health professionals, including interpretation of laboratory reports conveying complex genetic results and data, both pathogenic mutation and those of uncertain significance;
- A service that meets the needs of patients and their families, as monitored through validated patient satisfaction surveys.

2.3 Service Model

2.3.1 Clinical Genetics

Clinical Genetics will provide diagnostic and genetic counselling services, and in some multi-system disorders, co-ordination of management and follow up for individuals and families with, or at risk of, conditions which have, or may have, a genetic basis. Individuals and families should be helped to understand their condition, its implications, and their options with regard to reproduction, screening, prevention and management. The provision of advice on clinical management of patients with a genetic condition, and their wider family, is a key component of the role of Clinical Geneticists.
Clinical Genetic services will be predominantly outpatient based.

Each service will include:

- Diagnosis or exclusion of genetic disorders and congenital malformations;
- Investigation and genetic risk assessment;
- Provision of information;
- Predictive genetic testing;
- Discussion of reproductive options;
- Initiation and coordination of health surveillance and screening for genetic conditions;
- Co-ordination of interventional management in specialist or multi- disciplinary clinics;
- Management of the extended family;
- Maintenance of genetic family disease specific records;
- Liaison with genetic laboratories;
- Participation in local and national genetic networks;
- Education and training of genetic and other healthcare professionals.

Acting as an expert resource to all health professionals

- Audit of clinical services ;
- Research – clinical, biomedical, psychosocial and service related
- (Developed from 'Roles of the Clinical Geneticist' 2011, Clinical Genetics Society);

Service outputs will include:

- Clinical and genetic diagnosis, explanation and information about the disease, syndrome or condition, determination and communication of genetic/recurrence risk,

identification of screening and/or intervention options and appropriate counselling support;
- Service planning for integrating molecular testing in pathology and clinical genetics into mainstream specialities;
- The provision of expert information and educational resources to healthcare disciplines;
- Contribution to research through clinical and laboratory projects and recruitment to national studies and therapeutic trials;
- Liaison with colleagues: Clinical Geneticists and genetic counsellors will support colleagues across the medical specialties in order to ensure that the potential benefits and limitations of genetic testing are understood, made available to patients in all areas of medicine (referred to as 'mainstreaming'), and ethically applied. The clinical genetics services often provide an important 'gate-keeping' function with respect to optimising the utility of genetic tests. Examples of effective working arrangements include clinical networks, multidisciplinary teams and joint clinics. The liaison function includes ward consults and ward rounds;
- For some multi-system disorders with a genetic basis, Clinical Geneticists will provide on-going co-ordination of their management and follow up, linking with other specialties as appropriate. The majority of patients will either be discharged on receipt of diagnosis /advice or discharged back to the care of their referring clinician for ongoing management of their condition.

Genetics counsellors (GCs) should have appropriate professional registration. Regardless of location they will be professionally responsible to a Lead Genetic counsellor based in a regional genetic centre. GCs manage a proportion of genetic referrals under varying levels of supervision depending upon competency, experience and professional registration. The GC profession is currently undergoing statutory regulation led by the Association of Genetic Nurses and Counsellors (AGNC). The Genetic Counsellor Registration Board (GCRB) currently oversees professional registration. When in place, it will be the responsibility of the clinical genetics provider to ensure clinicians met the minimum requirements for registration and remaining on the register.

2.3.2 Laboratory Genetic Services

All requests for genetic testing will be through the All Wales Medical Genetic Centre at Cardiff, if the test is not available within the Centre, it will be sent to a Regional Genetics Centre within the UKGTN network, or to an accredited service worldwide.

Laboratory genetic services for the population of Wales will include:

- Genetic testing and clinical interpretation for:
  - Confirmation or exclusion of diagnosis;
  - Predictive and confirmatory testing in at-risk family members;
  - Prenatal testing;
  - Prenatal screening;
  - Carrier testing;
  - Precision medicine to determine prognosis, or prediction of treatment;
  - Support for clinical trials and research projects;
  - Long term banking of DNA (under the terms of the Human Tissue Act);
- Provide an expert resource for referring healthcare professionals.
- Co-ordination of investigations (including pre-natal diagnosis).
- Maintain Laboratory quality management and accreditation;
- Including Audit, training, quality assurance, equipment maintenance, staff competence, incident reporting.
- Contribute clinically significant genetic variant information to appropriate data repositories.
- Research and development;
  - Introduction of new technologies;
  - Validation and Introduction of new services;
  - Development and delivery of translational research projects.
- Education & training: of staff, trainees, other healthcare professionals, public.
- Attendance and contribution to MDTs.

In addition the service will be required to:

- Contribute to Welsh and UK genetic policy through any mechanisms as agreed by the WHSSC Medical Director to provide general advice to WG and WHSSC.
- Determine and implement the most clinically and cost effective means of delivering genetic and genomic services (e.g. targeted analysis/gene panels/sequential tests).
- Service planning for integrating molecular testing in pathology and clinical genetics into mainstream specialities.
- Liaison with colleagues: Clinical Scientists support colleagues across the medical specialties in order to ensure that the potential benefits and limitations of genetic testing are understood, made available to patients in all areas of medicine (referred to as 'mainstreaming').
- Participate in local and national genetic networks.

The laboratory service will have systems in place for managing test requests, which allow them to be processed in a clinically appropriate, cost effective and timely way, in accordance with the testing turnaround times, Association of Clinical Geneticists Guideline 2015, (see section 3.2.1). Laboratories will determine the most effective testing approach to be used, if necessary in discussion with the referrer.

Genetics laboratories must hold Clinical Pathology Accreditation (CPA)/ United Kingdom Accreditation Services (UKAS) and should provide accurate test results in accordance with nationally agreed target reporting times appropriate for the nature of the sample and referral reason.

If a request is made to the genetic laboratory by a Health Board/ NHS Trust which is not commissioned by Welsh Health Specialist Services the laboratory will need to advise the referrer the tests is not available within the current contract and will need to seek agreement for payment from the referring organisation.

2.3.2.1 *Laboratory Genetic Services Strategic planning*
Providers of laboratory genetics testing will also:

- Advise and support referrers on appropriate tests to carry out, including any mechanisms as agreed by the WHSSC Medical Director to provide general advice to WG and WHSSC.;
- Provide clinical interpretation of results;

- Determine the most clinically and cost effective means of carrying out testing (e.g. targeted analysis/gene panels/sequential tests);
    - Service planning for integrating molecular testing in pathology and clinical genetics into mainstream specialities;
    - Liaison with colleagues: Clinical Scientists support colleagues across the medical specialties in order to ensure that the potential benefits and limitations of genetic testing are understood, made available to patients in all areas of medicine (referred to as 'mainstreaming');
    - Contribution to research and service development through clinical-laboratory projects;
- Contribute clinically significant genetic variant information to appropriate data repositories;
- Maintain laboratory accreditation and quality service provision;
- Participate in local and national genetic networks;
- Educate and train genetic and other healthcare professionals;
- Act as an expert resource to all health professionals;
- Carry out audits of laboratory services;
- Carry out research - clinical, biomedical, and service related.

2.4 Service Delivery

2.4.1 Clinical Genetics

Genetic Services are available for those individuals (the affected individual), and their families, affected by, or concerned about, a disorder with a significant genetic component.

Patients can access clinical genetics via referrals from the following areas:

a) Primary Care i.e. GPs or other healthcare professionals
b) Secondary Care i.e. Consultants/Services
c) Tertiary Care i.e. Specialist Services
d) Self/ Family i.e. Family of existing AWMGS patients

Self-referrals are received from individuals and families that are already known to the service and 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 patients' family.

Referral to clinical genetics services may be indicated when an inherited disorder is identified or suspected, for example,

- Paediatrics including Dysmorphology;
- Specialist clinics: e.g. Rett syndrome, Tuberous Sclerosis;
- Cancer Genetics;
- Cardiovascular genetics: referral criteria under development;
- Prenatal Genetics;
- Neuromuscular Genetics;
- Endocrine genetics and disorders of sexual development;
- Familial Hypercholesterolaemia;
- Huntington's Disease;
- Neurogenetics.

The patient contact will be primarily outpatient-based, with clinics provided across Wales.

Background information should be gathered prior to a patient being seen as this can help to determine whether an appointment is required. Background information may include a detailed family history, confirmation of diagnoses (e.g. from cancer registry), review of medical records, psychosocial circumstances and, in some cases, preliminary genetic tests e.g. array-based comparative genomic hybridization (aCGH), fragile (X).

A proportion of the referrals should be managed by Genetic Counsellors under varying levels of supervision depending upon competency, experience and professional registration.
Services will provide both general and specialist clinics, e.g. paediatric, cancer, neurogenetics, prenatal and cardiac as well as some combined clinics e.g. ophthalmology, skeletal dysplasia, cardiac, dermatology, foetal medicine. (In some circumstances it is beneficial for patients thought to have a genetic condition to attend a single multidisciplinary clinic where medical/surgical management can be discussed and clinical genetics expertise is available. (The funding for the joint clinic will come via whichever service the clinic is recorded under.)

Outpatient 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.

Clinical genetic services requesting tests from laboratories should monitor requests for compliance with referral criteria stipulated by the UKGTN (included in the UKGTN NHS Directory of Genetic Testing, as this promotes efficient use of resources) or WHSSC Specialised service Commissioning Policies.

Inpatient work is usually urgent and includes ward consultations and prenatal advice in foetal medicine units. The Regional Genetics centres 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 their region. They should also offer ward consults (including neonatal and intensive care units) and rapid access clinics for urgent prenatal queries in their host trusts and where possible, or volume requires, in linked district general hospitals (DGHs) too, depending on location.

2.4.2 Laboratory

There is a core list of genetic tests which the preferred laboratory must provide for the population of Wales, annex i. Core services includes both inherited (germline) and acquired (for cancer) tests.

2.4.2.1 Inherited services

Providers will be a member of the UK Genetic Network, UKGTN. In addition to providing core services, the laboratory will provide some specialist (UKGTN-recommended) services. (It should be noted that UKGTN covers inherited / germline tests only)

It is not anticipated that the provider will provide every test, therefore, they must have appropriate systems in place for transporting specimens to other laboratories (or advising referrers of referral routes for specialised tests) and receiving results back safely and relaying these to the requesting clinician.

All requests for UKGTN tests will be through the provider provider, who will direct to the relevant laboratory. It is the responsibility of the preferred provider to ensure results are received back in line with National reporting times and relayed to relevant clinicians.

UKGTN recommend tests annually for commissioning, currently, not all the UKGTN recommended tests are commissioned by WHSSC as they become available. As an interim measure between tests being recommended and tests being commissioned, the preferred provider will need to request funding via the IPFR process.

Furthermore, recommendations from the UKGTN for commissioning new tests will be considered by WHSSC as part of the Integrated Commissioning Planning process. A clinical geneticist and genetic scientist will advise WHSSC in relation to UKGTN recommended tests.

2.4.2.2 Genetic Testing for Stratified Medicine/ Treatment

The provider is commissioned to provide molecular services for acquired disorders; these are divided into solid tumours and the haematological malignancies. These services are not included on UKGTN. (See also Annex i)

All requests from referrers for genetic testing for new services for biomarkers to inform stratified medicine will be referred through to the genetic testing for stratified medicine Task and finish group. This includes any genetic testing recommended by NICE for stratification of medicine/ treatment. The provider will nominate a clinical scientist to be a full member of this group. If on reviewing the evidence there remain outstanding questions in relation to the evidence of benefit to impact on the clinical outcome/ cost effectiveness, the test will not be commissioned by WHSSC. However, a Health Board may take the decision to fund the test themselves as an interim measures whilst the review is taking place and if a decision is made by WHSSC not to fund.

Furthermore, it is recognised that laboratories will provide some genetic testing for stratification of medicine/ treatment to Welsh NHS patients through externally funded sources i.e. drug funded research.

In these situations the laboratory are wholly responsible for ensuring appropriate governance arrangements are in place. In addition, the laboratory will advise commissioners of the duration and funding implications for NHS Wales on entering any agreement.

Where genetic testing is available to NHS Wales as a result of a trial or external funding, the laboratory will need to ensure all

referring clinicians, for the specific test, are aware of the duration of the availability of the test. Once funding has ceased a full business case will need to be submitted through the provider/ referring Health Boards to WHSSC for consideration in the next available planning round.

As an interim measure between external funding for tests ceasing and tests being commissioned, the referring clinician will need to request funding from their Health Board IPFR team.

It is recognised that genetic laboratories will also provide other tests, outside of the WHSSC contract for example:

- During development of relevant tests prior to approval by the UK GTN;
- For acquired genetic conditions, for example genetic testing of sporadic cancers;
- For external contracts for non-Welsh referrers;
- For externally funded research projects.

Whilst Welsh NHS patients may have access to these, they are not WHSSC funded and the laboratory will need to ensure suitable governance arrangements are in place.

It is recognised that advances in genomic medicine and the supporting technologies are leading to the identification of new genetic conditions and new methods of testing/ diagnosis. Tests developed in the laboratories using new technologies need to be validated for clinical diagnostic use (e.g. panel tests that use Next Generation Sequencing (NGS). In addition, changes in testing methodologies used by the laboratory will require the development of business case which will need to be supported by the provider and presented to commissioner of genetic services, for consideration through WHSSC planning process.

2.4.2.1 Laboratory Genetic Services for Nationally Defined Specialised Services

Services provided by specialised genetic laboratories for nationally defined specialised services may include:

- Diagnostic testing for children with learning difficulty, dysmorphism, developmental anomalies or symptoms indicative of a constitutional or acquired chromosomal anomalies or singe gene disorder.
- Diagnostic testing in children or adults showing signs and symptoms that may be indicative of a genetic cause.

- Germ line mutation identification in both common (e.g. breast, bowel) and rare cancers and some common adult disorders, for example cardiac, endocrine, renal and neurological conditions where there is a significant family history in addition to symptoms in the index case. This is with the intention of offering confirmatory testing and pre-symptomatic testing in relatives of index cases.
- Pre-symptomatic testing in adults at high risk of some late onset inherited conditions including Huntington disease and familial cancers.
- Specialised services for children at high risk of a condition where management intervention is required in childhood, e.g. retinoblastoma or Multiple Endocrine Neoplasia (MEN) and related cancer syndromes.
- Definitive prenatal testing for pregnant women in whom screening identifies an increased risk of constitutional chromosomal imbalances and abnormalities e.g. Downs syndrome and related aneuploidies.
- Prenatal testing for single gene disorders e.g. cystic fibrosis and Duchene muscular dystrophy.
- Testing for chromosomal imbalances and other genetic abnormalities indicated by reproductive history e.g. cases of recurrent miscarriages and male infertility.
- Carrier testing in adults at risk of an adverse reproductive outcome from balanced chromosomal conditions and some single gene conditions e.g. congenital adrenal hyperplasia.
- Testing to confirm an abnormal result from a population screening programme (funding of confirmatory tests is included as part of the diagnosis/treatment of the index patient) Services should ensure that there are links in place with population screening programmes.
- Long term banking of cells, preserved material and DNA (under the terms of the Human Tissue Act) to facilitate the long term commitment of medical genetic services to validate, provide and quality assure diagnostic and counselling services to families and future generations at risk of inherited disorders and to contribute to medical research under ethical committee approval.
- Molecular Pathology tests to inform genetic testing (e.g. Immunohistochemistry and microsatellite instability).

2.4.3 Cancer Genetic Services

Although 1 in 2 of the population is expected to develop a cancer, only 5-10% of cancers of the breast, ovary and colon are due to an inherited predisposition and require testing from laboratory services.

Referral guidelines to the cancer genetics service can be found at:
http://www.wales.nhs.uk/sites3/documents/966/GENETICS_REFERRAL_GUIDELINES_1.pdf

Currently the Cancer referral criteria within the preferred provider are being reviewed and updated to allow for new NICE guidance (www.nice.org.uk/guidance/cg164) and GUT guidelines (www.ncbi.nlm.nih.gov/pubmed/23408351).

2.5 Tertiary referral management for genetic testing / Gate-keeping

The preferred provider will provide clinical advice and gate-keeping in relation to the following areas

2.5.1 Cardiogenetics-CP57

The Inherited Cardiac Conditions Multi Disciplinary Teams, ICC MDTs, are the gatekeepers for referrals to the All Wales Medical Genetic Service for cardiogenetic testing. All referrals to the AWMGS for cardiogenetic testing must be approved by the regional ICC MDT. Referrals that have not been approved by the ICC MDT will be returned to the referrer. There are 3 regional ICC MDTs: North Wales, South West Wales and South East Wales.

2.5.2 Preimplantation Genetic diagnosis –CP37

For further detail on PGD, the most up to date policy can be found at
http://www.whssc.wales.nhs.uk/policies-and-procedures

2.5.3 UKGTN Genetic Tests

Access to UKGTN tests is detailed in section 2.4.2.

2.6 Care Pathway

Given the diversity of the diseases seen by medical genetics, there is no generic pathway for access into/ through the services. Instead, the service has a number of disease based clinical pathways available on the clinical genetics website

http://www.wales.nhs.uk/sites3/page.cfm?orgid=525&pid=17830

3. QUALITY AND PATIENT SAFETY

3.1 Quality and Patient Safety

Patients/families must 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 or telephone consultations summarised in writing afterwards.

- 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.
- Consent for genetic testing and retention of deoxyribonucleic acid (DNA) samples must be undertaken in accordance with Department of Health guidance and the Mental Capacity Act, 2005.
- Education is provided at clinic, through post-clinic letters and in patient information leaflets (in different languages).
- 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 neuromuscular disorders. Advocacy and support should be offered to carers appropriately.

The Provider must work to written quality standards, as detailed in the genetic dashboard, annex ii and provide monitoring information to the lead purchaser.

The centre must enable the patient's, carer's and advocate's informed participation and to be able to demonstrate this. Provision should be made for patients with communication difficulties and for children.

3.1.1 Laboratory required testing report times

Accurate test results provided in accordance with the agreed reporting times. Reporting times vary depending on the nature and urgency of the referral (potential diagnosis, sample type, scale of testing, prenatal tests, etc). Typical target report times in current professional standards guidance are:

Reporting Time Target (calendar days)	**Definitions**
3 days	Rapid aneuploidy QF-PCR/PCR/FISH testing for prenatal, postnatal or oncology referrals
	Rapid PCR/FISH testing for haemato-oncology referrals
	PCR-based tests where the result is needed urgently for prenatal diagnosis
10 days	Karyotype for urgent postnatal blood referrals
14 days	Karyotype results for prenatal referrals
	Karyotype results for urgent haemato-oncology referrals*
	Array CGH results for prenatal referrals and urgent postnatal
	Southern blot tests where the result is needed urgently for prenatal diagnosis
	PCR-based tests for predictive testing and confirmation of neonatal results.
21 days	Karyotype results for routine haemato-oncology referrals
28 days	Karyotype results for routine postnatal blood referrals
	Routine testing of solid tissue referrals
	Array CGH results for postnatal referrals
	Non-urgent PCR-based tests where the familial mutation is known (excluding predictive tests), specific mutation tests, or gene tracking by microsatellite analysis
56 days	Mutation screening or tests that require Southern blot analysis
	Next generation sequencing panels of ≤10 genes
112 days**	Next generation sequencing panels of >10 genes** and other large scale sequencing work e.g. WES and WGS

SOURCE: Association of Clinical Genetics Scientists Guideline (2015)

It is recognised in order to improve the quality of the laboratory services, the laboratory may seek capital funding through other available public funding streams i.e. Welsh Government technologies fund for equipment. In these instances the laboratory will need to ensure, WHSSC are aware of the request for funding and the implications for revenue funding if the bid is successful, in order to consider through the NHS planning cycle.

3.2 Quality Indicators (Standards)

Core standards are:

- Familial Breast Cancer, 2006. NICE clinical guideline 41 (partial update) www.nice.org.uk
- Standards for medical laboratories, 2010 Clinical Pathology Accreditation – www.cpa-uk.co.uk
- National waiting times and access criteria as outlined in the National Operating Framework.
- The UK GTN provides information relevant to genetic diagnosis for rare conditions on quality and standards for genetic testing and a Directory of Genetic Testing that lists tests that have been recommended for NHS service and approved for clinical validity and utility – www.ukgtn.nhs.uk
- Recommended standards are set out in the Best practice Guidelines of the organisations listed below (and detailed in section 1 Evidence Base).
- The British Society for Genetic Medicine – www.bsgm.org.uk and Association for Clinical Genetic Scientists- www.acgs.org.uk

3.3 Putting Things Right: 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:

- When a patient or their representative is unhappy with the decision of the gatekeeper, that the patient does not meet the criteria for treatment and that the patient is not an exceptional case, the patient and/or their representative has a right to ask

for this decision to be reviewed. The review should be undertaken, by the patient's Local Health Board, in line with section 7 of the All Wales Policy: Making Decisions on Individual Patient Funding Requests;

- When 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.

4. PERFORMANCE MONITORING AND INFORMATION REQUIREMENTS

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.

WHSSC and the service will meet on a quarterly basis to review performance of the service against the service specification. However, the service will formally inform Director of Planning, WHSSC immediately if they have concerns they will not be able to meet the requirements within the service specification. They will also advise what remedial action is proposed to ensure they meet the requirements of the service specification.

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

4.1.2 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).

Proxy outcome measures that will be monitored and for which providers will be required to submit information, for both clinical geneticist and genetic counsellors are:

- New to follow up ratios.
- Numbers of new and follow up patients seen.
- Proportion of appointments held at RGC compared with proportion in outreach clinics.
- Reason for referral broken down into main disease categories (this will aid future service planning).
- Percentage of ethnic origin data recorded.
- Numbers and type of tests sent to other laboratories.

The cardiogenetics services will provide an annual report in line with the report requirements detailed within CP57 Cardiogenetic policy. In addition to the annual report for the service, data from the cardiogenetics service will be presented by a member of the ICC MDTs at the annual Cardiac Audit Day held in November each year**.**

The FH service will also provide an annual report to include the following details.

- Number of index tests undertaken: trajectory of predicted levels of testing.
- Number of family cascade tests undertaken: trajectory of predicted levels of testing.
- Percentage positive index cases.
- Average number of cascade tests per index case.
- Financial position.
- Waiting list position.
-

4.1.3 Laboratory Genetics

- The following will be reported to WHSSC by the AWMGS annually, on a quarterly basis for the core services:
- Number of samples received.
- Number of samples extracted.
- Number and type of samples exported.
- GenU activity (separated into MolU and CytU).
- Financial position.
- Compliance with DH waiting times for lab.
- Serious incidents regarding lab tests - number of serious incidents involving laboratory tests.

In relation to BRCA testing

- Number of patients tested between 10 and 20% risk.
- Number of patients tested with a risk greater than 20% risk.
- Percentage of positive index cases.
- Number of family members taking out of screening as a result of predictive testing.
- Reporting times (screening and predictive testing).
- Financial position – this will be difficult as the budget will be combined with the lab and clinical budgets.
- Waiting list position – for clinical.

External Quality Assurance (EQA) - scores from EQA schemes the laboratory participates in

- Activity audits (laboratory) - proportion of nationally agreed audits participated in by the genetics laboratory (the number and type of audits need to be agreed)

The summary measures are listed below:

- Pick up rate for genetic testing - Proportion of tests that return a positive result for affected patients that have the test to determine a diagnosis and are seen in clinical genetics.
- Multi Disciplinary Clinics (MDC) - Proportion of clinical genetic clinics that are part of a MDC/multi-disciplinary team (MDT).
- 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).
- 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.
- Patients waiting excessively for pre-natal (PN) genetic test results where the proportion of patients receiving test result within 5 working days after the clinic receives the laboratory report for PN genetic test results.
- Do Not Attends (DNAs) as defined in the Data Dictionary - proportion of appointments that are not attended.

- Patients consulted without a referral.
- 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.
- All serious incidents will be reported within two days of incident.
- Serious incidents regarding patient care.
- Serious incidents regarding laboratory tests.
- Non-adherence to the UKGTN Testing Criteria (laboratory) as per UKGTN website - proportion of test requests from clinical genetics that do not comply to the UKGTN Testing Criteria where those criteria apply.

4.2 Key Performance Indicators

The providers will be expected to monitor against the genetic dashboard measures, annex ii.

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 Any acceptance and exclusion criteria

Referral processes and sources

Clinical Genetics referrals:

- Referrals will be accepted from primary, secondary and tertiary care.
- Referrals may originate from all areas of medicine, most commonly general practice, paediatrics, obstetrics/antenatal screening services, oncology, neurology, surgery, cardiology, dermatology, nephrology, and audiology.
- Referrals may be urgent, e.g. neonatal or ward consultations, or for urgent prenatal advice and may be received from other Regional Genetics Centres (RGCs) if families are widely scattered. 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 General Practitioner 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.

Criteria for a referral

- Detailed referral guidelines for clinical referrals and referral criteria for joint/multidisciplinary clinics should be provided on RGC websites.
- Clinical Genetics services should have robust referral criteria in place to ensure that they only see patients for whom they are likely to be able to offer advice.
- Criteria for consultant to consultant referrals to Clinical Genetics Services are under development.
- 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 seen in a clinic.

Laboratory service referrals

- Only requests from Clinical Geneticists/ hospital consultants and appropriately registered Genetic Counsellors working for Regional Genetic Centres will fall within the WHSSC contract. (AWMGS Laboratory will ensure necessary billing for tests ordered by other clinicians)

Criteria for a referral

- 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 UK GTN testing criteria, where these are available. For acquired disorders, the appropriate clinical referral guidelines should be followed. Where the UK GTN's testing criteria are available these should be adhered to in the ordering of tests.
- Tests not approved by the UKGTN are excluded from this specification, with the exception of rare tests accessed from abroad which are not available in the UK. The UKGTN Directory includes validated molecular and cytogenetic tests (including agreed 'grandfather' tests developed prior to the UKGTN.)

Referral handling

- Laboratories will have systems in place for managing test requests, which allow them to be processed in a clinically

appropriate, cost effective and timely way, in accordance with the testing turnaround times (see section 3.2). Laboratories will determine the most effective testing approach to be used, if necessary in discussion with the referrer.

- The preferred provider will not provide every test internally, thus, the laboratory will have an appropriate systems in place for transporting specimens to other laboratories and receiving results back safely and relaying these to the requesting clinician.
- The following exclusions apply:
- Patients requiring specialist procedures should be referred to the appropriate specialists
- Genetic testing within population screening programmes which are not detailed in annex 2 core services.

4.4 Referral Pathway

The service have a number of disease based clinical pathways available on the clinical genetics website

http://www.wales.nhs.uk/sites3/page.cfm?orgid=525&pid=1783

5. EQUALITY IMPACT AND ASSESSMENT

The Equality Impact Assessment (EQIA) 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 Equality Impact Assessment. The Assessment has shown that there will be no impact.

Annex i Core Services Commissioned through WHSSC

Gene analysis	Service type	Notes
Amniotic fluid	Inherited	
Chorionic villi	Inherited	
Blood – karyotyping	Inherited	
Haematological malignancy (AML, ALL, CML, CLL, MPN)	Acquired	Mixed funding – some requests funded through HB SLAs
Solid tissues	Inherited	
array CGH (LD)	Inherited	1025 contracted samples pa
FISH	Inherited	600 contracted samples pa
QF-PCR	Inherited	~1500 contracted samples pa
FISH send-outs	Inherited	Funded through genetic send outs
AS/PWS	Inherited	Analysis through SCOBEC
BRCA screening	Inherited	590 contracted samples pa
BRCA PSTs	Inherited	
CF	Inherited	Includes newborn screening
DM	Inherited	
DMD/BMD	Inherited	
DRPLA	Inherited	Analysis through SCOBEC
EGFR	Acquired	350 contracted samples pa
FAP screens and PSTs	Inherited	
Familial hypercholesterolaemia screens and PSTs	Inherited	250 contracted samples pa (budget held by Clinical Genetics)
FraX	Inherited	
FRDA	Inherited	Analysis through SCOBEC
HD	Inherited	
HMSN/HNPP	Inherited	
HNPCC screens and PSTs	Inherited	120 contracted samples pa
LHON	Inherited	
SCA 1, 2, 3, 6, 7 & 17	Inherited	
SMA	Inherited	
XBSMA	Inherited	Analysis through SCOBEC
Banked DNA	Inherited	
Send outs	Inherited	Funded for £50k (WHSSC) plus £170k (C&V UHB)

Annex ii Quality Dash Board

Dashboard measures

The summary measures are listed below.

- Pick up rate for genetic testing - Proportion of tests that return a positive result for affected patients that have the test to determine a diagnosis and are seen in clinical genetics
- Multi Disciplinary Clinics (MDC) - Proportion of clinical genetic clinics that are part of a MDC/multi-disciplinary team (MDT)
- 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)
- Laboratory reporting times - Proportion of Cytogenetics reports meeting turn round times as agreed by the professional organisations (CMGS/ACC)
- Laboratory reporting times - Proportion of Molecular reports meeting turn round times as agreed by the professional organisations (CMGS/ACC)
- 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
- Poor patient experience - number of written complaints about the genetics department
- Good patient experience - number of letters/emails from patients, carers or non-genetics consultants registering thanks to the genetics department
- Patients waiting excessively for pre-natal (PN) genetic test results where the patient is seen in the clinical genetics department - proportion of patients receiving test result within 5 working days after the clinic receives the laboratory report for PN genetic test results.
- Do Not Attends (DNAs) as defined in the Data Dictionary - proportion of appointments that are not attended
- Patients consulted without a referral
- 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
- Serious incidents regarding patient care - number of serious incidents involving patient care
- Serious incidents regarding lab tests - number of serious incidents involving laboratory tests

- External Quality Assurance (EQA) - scores from EQA schemes the laboratory participates in
- Activity audits (laboratory) - proportion of nationally agreed audits participated in by the genetics laboratory (the number and type of audits need to be agreed)
- •Non-adherence to the UKGTN Testing Criteria (laboratory) as per UKGTN website - proportion of test requests from clinical genetics that did not comply to the UKGTN Testing Criteria where those criteria apply

Genetics Dashboard

Reference	Category	Examples of tests to be included in category
GEN04i	Proportion of prenatal and urgent postnatal rapid aneuploidy QF-PCR/FISH tests and PCR based tests where result is required urgently for prenatal diagnosis completed within 3 working days.	Prenatal test on DNA from CVS, AF, fetal blood (including rapid aneuploidy) by PCR, Sanger sequencing, FISH or chromosome analysis from direct CVS culture. Neonates with query Trisomy (FISH).
GEN04ii	Proportion of urgent haemato-oncology rapid PCR/FISH tests completed within 3 working days.	Urgent diagnostic haemato-oncology and molecular monitoring of acute leukaemias.
GEN04iii	Proportion of urgent postnatal blood karyotype tests completed within 10 calendar days.	Urgent postnatal blood karyotype and urgent chromosome breakage.
GEN04iv	Proportion of prenatal cytogenetics tests & urgent postnatal array CGH tests, PCR based tests for predictive testing and confirmation of neonatal results, and Southern blot analysis where the result is urgently needed for prenatal diagnosis completed within 14 calendar days.	Prenatal chromosome analysis, prenatal microarray, prenatal FISH and prenatal Southern blots (FSHD, Fragile X syndrome). Clinically urgent PCR, MLPA, Sanger sequencing, microarray and FISH tests (not prenatal); neonatal diagnosis or patient/partner pregnant. BRCA predictive tests. Predictive testing PCR, MLPA or Sanger sequencing where familial pathogenic mutation is known.
GEN04v	Proportion of urgent haemato-oncology tests completed within 14 calendar days.	Karyotyping for ? acutes, ? CML ? transformation and ? relapse.
GEN04vi	Proportion of routine haemato-oncology tests completed within 21 calendar days.	All non-urgent referrals.
GEN04vii	Proportion of routine postnatal/solid tissue/bloods cytogenetic tests including array CGH tests, non-urgent PCR based tests where the familial mutation is known (excluding predictive tests) and specific mutation tests or gene tracking by microsatellite analysis completed within 28 calendar days.	Routine postnatal karyotype. Routine and solid tissue microarrays. Non-urgent microarray follow up tests. Chromosome breakage testing. Diagnostic and cascade testing by PCR, MLPA, Real time PCR. Methylation pyrosequencing analysis for FSHD2. Fragile X Assuragen test. Cascade Sanger sequencing of known variants if not urgent. Cascade MLPA or Sanger sequencing for familial segregation analysis of an unclassified variant.
GEN04viii	Proportion of mutation screening or tests that require Southern blot analysis and Next Generation Sequencing panels (NGS) of ≤10 genes completed within 56 calendar days.	Full (or partial) gene screen by Sanger sequencing. Small NGS panels of ≤10 genes. Southern blotting (e.g. FSHD and Fragile X syndrome).
GEN04ix	Proportion of Next Generation Sequencing (NGS) panels of >10 genes and other large scale sequencing work e.g. WES or WGS completed within 112 calendar days.	Large targeted NGS panels of >10 genes. Clinical exome panels. Whole exome and whole genome sequencing