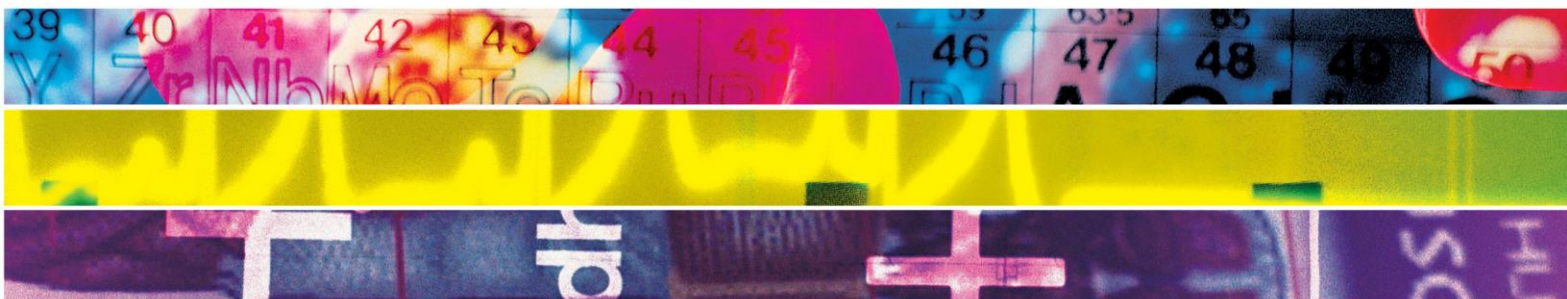


# National Healthcare Science School of Genetics

## Healthcare Science Practitioner Training Programme in Clinical Laboratory Genetics

### Training Manual

#### Modernising Scientific Careers



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## **1.0 Healthcare Science Practitioner Training Programme (PTP) in Genetics**

### **1.1 Introduction**

This manual describes the structure and the functions of the training programme for Healthcare Science Practitioners (HCSP) in Clinical Laboratory Genetics. The training programme has been developed under Modernising Scientific Careers (MSC) and funding from the Department of Health (DH) Genetics Unit. It is now the formal training programme for HCSP and is endorsed by the National Healthcare Science School of Genetics, the Genetics Education and Training Board of the Association for Clinical Cytogenetics (ACC) and the Clinical Molecular Genetics Society (CMGS) and the Association of Genetic Technologists (AGT). The document describes the training programme over the next two years, the assessment strategies and the portfolio structure.

Funding and support is provided through the Strategic Health Authorities (SHAs) who fund trainee placements.

### **1.2 Good Scientific Practice (GSP)\Good Laboratory Practice (GLP)**

The principles outlined in Good Scientific Practice\Good Laboratory Practice are the standards to which trainees will be expected to adhere and are fundamental to both PTP and STP.

Good Scientific Practice sets out, for the profession and the public, the standards of behaviour and practice that must be achieved and maintained in the delivery of work activities and care provided.

Good Scientific Practice aims to contextualise and make more explicit the standards of practice and proficiency set down by the Health Professions Council (HPC) in a way that is accessible to the profession and the public. It uses as its basis the HPC Standards of Proficiency and HPC Standards of Conduct, Performance and Ethics, but elaborates these within the context of the modalities within the sphere of Healthcare Science.

The Domains of Good Scientific Practice are,

1. Professional
2. Scientific
3. Clinical
4. Technical
5. Investigation and Reporting
6. Quality
7. Working with colleagues
8. Research and development
9. Probity
10. Leadership
11. Training and Developing Others

**Appendix 1** sets out the details of Good Scientific Practice.

Reference to Good Laboratory Practice can be found on the Clinical Pathology Accreditation (CPA) website. Reference to Good Clinical Practice can be found on the Medicines and Healthcare Products Regulatory Agency (MHRA) website.

### **1.3 Assessment**

The assessment tools for both the Practitioner and Scientist training courses are integral to successful completion of the training programme. The assessments are designed to test a range of skills and competencies in line with the agreed curriculum for the programme and must be successfully completed using the online system provided.

The electronic portfolio is designed for both Practitioner and Scientist training programmes. It provides support to the trainees with their continuous professional development throughout the training programme and provides a mechanism through which their development and progress can be monitored and managed.

The National Healthcare Science School of Genetics has developed an operational manual to provide guidance on the use and management of the assessment tools “The Online Assessment and Personal Development Management System”.

Access to the online system and supporting documentation is available from the National Healthcare Science School of Genetics; [Genetics.nwd@westmidlands.nhs.uk](mailto:Genetics.nwd@westmidlands.nhs.uk).

### **1.4 The role of a Healthcare Science Practitioner (HCSP)**

It is important to consider the roles of the Healthcare Science Practitioner in relation to those working in other roles in the career framework.

- A HCSP in genetics will have technical expertise within genetics and will work in a range of healthcare settings related to genetics.
- HCSP are likely to work under indirect supervision but with a flexible approach to meet the needs of patients and of the service.
- Specific and extended roles developed for genetics practitioners are seen in **Appendix 2**.

## **2.0 The Training Programme**

### **2.1 Aim**

The training programme is designed to provide the HCSP with strong technology based training across all aspects of genetics with an appropriate level of underpinning knowledge to enable them to perform genetic analyses in a range of healthcare settings. The training will be predominantly work place based. In addition, it is planned that throughout the training

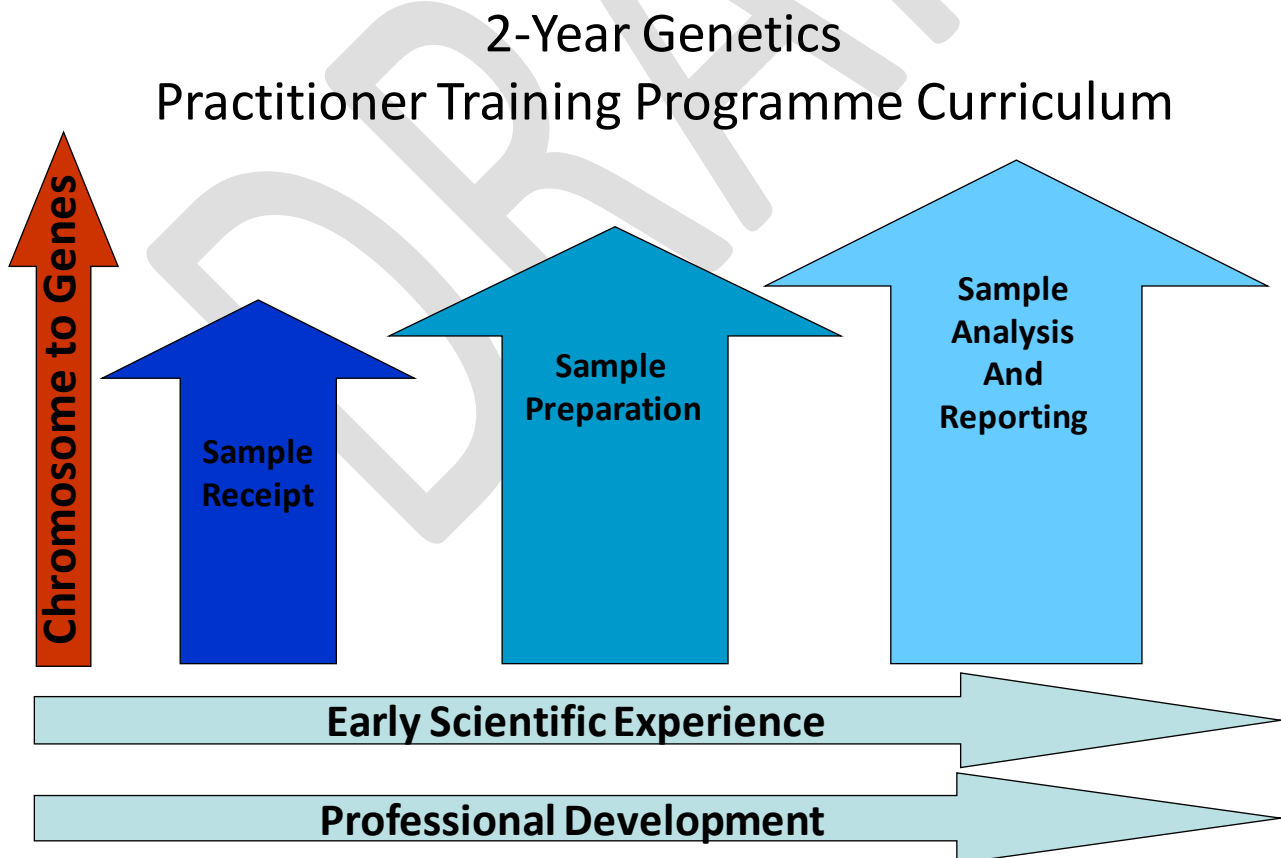
programme there will be additional training opportunities to enable trainees to spend time in a range of healthcare and industrial settings to provide experience in the most recent technological advances in genetics.

## 2.2 Module Design

The programme is designed to follow the patient's sample through the laboratory, such that the trainee HCSP acquires all of the required skills and competencies, from sample receipt and preparation to analysis, with the final aim being sample analysis and reporting the results of routine cases.

There is in addition, a generic module covering all aspects of professionalism, which will be spread over the two years of the programme, and themed through the work based training modules. The aim of the generic component of training is to prepare the trainee to undertake appropriate and effective leadership and managerial responsibilities and to take on appropriate patient responsibilities within the HCSP role. The indicative content of the generic programme includes an understanding and delivery of the standards contained in Good Scientific Practice/Good Laboratory Practice, including the development of a patient focus, leadership and management as appropriate to the role, developing an understanding of quality and clinical governance, health and safety and the context of science within the NHS.

**Figure 1 Healthcare Science Practitioner Training Programme Curriculum**



The training programme will begin in October in the host laboratory and should begin with an induction into the local laboratory.

This induction is laboratory based learning and should cover the following areas as well as anything else pertinent to the local laboratory:

- Local hospital induction – local policies
- Review of local service and functioning of the laboratory
- Review and more detailed description of health and safety, pertinent to the modules and to the local laboratory
- Detailed knowledge of laboratory policies relating to health and safety
- Basic knowledge of reagents and chemicals used within the laboratory including awareness of Control of Substances Hazardous to Health (COSHH), safe handling and risk assessments
- Basic knowledge of biological risks within the laboratory, and awareness of safety containment procedures for different risk categories
- Basic knowledge about the function, operation of routine laboratory equipment used throughout the department, e.g. safety cabinets, centrifuges etc.

### **2.3 Joint meeting with Healthcare Scientist (HCS) Trainees**

Early in the programme Healthcare Scientist and Healthcare Science Practitioner trainees will come together in a national course – organised by The National Healthcare Science School of Genetics. This is likely to include:

#### **2.3.1 Indicative Content of Course**

- Introduction to MSC
  - Career development framework
  - Exciting developments across healthcare
- Overview of course and expectations
  - Roles of HCSA, HCSP and HCS within the laboratory
  - Course design and structure
  - Assessment overview
  - Introduction to health service values and structures
  - Equity and Excellence: Liberating the NHS – current philosophies of patient care
  - House of Lords Review of Genomic Medicine
- Current and future role of Healthcare Scientists and Practitioners
- Introduction to ‘Good Laboratory Practice’ and ‘Good Scientific Practice’
  - Professionalism
  - Consent level and confidentiality
  - Legal frameworks
  - Clinical governance
  - The role of audit and research
- Introduction to health and safety
  - Basic laboratory H&S
  - Safe systems and processes

- Laboratory behaviour
- Importance of samples / patient pathways
- Introduction to basic quality control
  - National External Quality Assurance System (NEQAS)
  - Quality assurance
  - Laboratory accreditation
  - Role of performance management in maintaining quality
  - Role of innovation and service improvement
- Bringing technology into healthcare
  - Sequencing
  - Automation
  - Bioinformatics
  - Examples from other disciplines
- Organisation of Genetic Services in the UK
  - Laboratory and Clinical Genetic Services
  - Professional bodies (CMGS/ACC and BSHG)
  - UK Genetic Testing Network (UKGTN)
  - National Healthcare Science School of Genetics

## 2.4 Key Principles

There are a number of principles within all of the following modules;

- 1 Understanding the key quality parameters
- 2 Being competent in the technique such that those quality parameters are achieved consistently and
- 3 Understanding the clinical context and the limitations of the test.
- 4 Understanding of Clinical Pathology Accreditation (CPA) and its requirements and adhering to these principles
- 5 Understanding of the role of standard operating procedures (SOPs).
- 6 Health and Safety
- 7 Understanding equipment use and maintenance

While these are specifically stated for some learning outcomes, although not all, it is understood that they **apply to all**.

## 2.5 MODULE 1 Sample Receipt and Preparation

This module has 6 learning outcomes. These are shown together to demonstrate the structure of the overall module and then the knowledge and competencies attached to each learning outcome are detailed individually.

### 2.5.1 Aim

To prepare the trainee to undertake safe receipt and handling of samples and prepare them for genetic analysis by application of the appropriate techniques.

## 2.5.2 Learning Outcomes

### Learning outcome 1

Demonstrate understanding and skills in the principles of safe receipt and handling of samples including data entry and storage.

### Learning outcome 2

Demonstrate understanding and skill in cell culturing, harvesting, slide making and staining techniques for chromosome analysis (including both short and long term culture).

### Learning outcome 3

Demonstrate understanding and skill in DNA and RNA extraction from a range of patient sample types. Evaluate the importance of the quality and quantity required for subsequent analyses.

### Learning outcome 4

Understand the principles of and successfully process samples for array analysis using a variety of platforms such as, SNP, OLIGO and Resequencing arrays, depending on local availability.

### Learning outcome 5

Prepare and successfully process an appropriate range of fluorescence in situ hybridisation probes (FISH), for example, chromosome enumeration probes, locus specific probes and whole chromosome paints.

### Learning outcome 6

Demonstrate the ability to perform a range of PCR techniques both manually and using automated technology, including sample preparation and validation of the outcome of the reaction.

## 2.5.3 Detailed Content

### Learning Outcome 1

Demonstrate understanding and skills in the principles of safe receipt and handling of samples including data entry and storage.

#### Indicative content and practical skills to be delivered in the workplace

- Laboratory procedure for receipt of samples.
- Patient confidentiality and the Data Protection Act.
- Requirement for and use of patient consent
- Sample requirements for a variety of tests and the procedures to employ when the requirements are not met.
- Safe handling of all samples including recognising the hazards posed by high risk samples and the safe disposal of biological samples.
- Laboratory procedure for the storage of the range of samples, both prior and

subsequent to processing.

- Legislation and regulations covering the transport and packaging of samples.
- Use of the laboratory information systems and other patient databases in relation to the receipt and storage of samples.
- Equipment use and maintenance.

## **Learning Outcome 2**

Demonstrate understanding and skill in cell culturing, harvesting, slide making and staining techniques for chromosome analysis (including both short and long term culture).

### **Indicative content and practical skills to be delivered in the workplace**

- Know and understand the cell cycle and how it can be artificially manipulated to maximise chromosome quality.
- Principles of cell culture and cell separation over a range of sample types.
- Principles and practice of aseptic technique.
- Structure and function of chromosomes.
- Cytogenetic slide preparation from a range of sample types.
- Principles of common staining and chromosome banding techniques and their appropriate use.

## **Learning Outcome 3**

Demonstrate understanding and skill in DNA and RNA extraction from a range of patient samples types. Evaluate the quality and quantity required for subsequent analyses.

### **Indicative content and practical skills to be delivered in the workplace**

- Principles of DNA and RNA extraction chemistries.
- Perform manual and automated methods of extracting DNA/RNA from a range of sample types, including knowledge of other automated extraction equipment and their limitations.
- Principles and methods available for determining DNA/RNA quantity and quality, and specific requirements for downstream processes.
- Ability to recognise and trouble-shoot problematic extractions.

## **Learning Outcome 4**

Understand the principles of and successfully process samples for array analysis using a variety of platforms such as, SNP, OLIGO and Resequencing arrays, depending on local availability.

### **Indicative content and practical skills to be delivered in the workplace**

- Parameters for adequate quality and quantity of extracted DNA for array work.
- All relevant quality parameters.

- Use and application of different array platforms.
- Principles and practice of sample processing to obtain results of diagnostic quality.

### **Learning Outcome 5**

Prepare and successfully process an appropriate range of fluorescence in situ hybridisation probes (FISH), for example, chromosome enumeration probes, locus specific probes and whole chromosome paints.

#### **Indicative content and practical skills to be delivered in the workplace**

- Principles of probe labelling and hybridisation.
- Understand the range of probes available and their common clinical application.
- Undertake fluorescence in situ hybridisation using a range of probes.
- Principles of epi-fluorescence microscopy.

### **Learning Outcome 6**

Demonstrate the ability to perform a range of PCR techniques both manually and using automated technology, including sample preparation and validation of the outcome of the reaction.

#### **Indicative content and practical skills to be delivered in the workplace**

- Understand the process of PCR and its clinical application.
- Understand the range of instrumentation available for PCR and their use.
- Principles of primer design and optimisation of reactions.
- Undertake manual and automated set up of PCR reactions.
- Principles of agarose gel electrophoresis
- Undertake fragment separation by gel electrophoresis.
- Parameters for adequate quality and quantity for amplified DNA and how to resolve problems.

## **2.6 MODULE 2 Sample Analysis – From Chromosomes to Genes**

This module has 6 learning outcomes. These are shown together to demonstrate the structure of the overall module and then the knowledge and competencies attached to each learning outcome are detailed individually.

### **2.6.1 Aim**

To prepare the trainee to accurately carry out a comprehensive range of genetic analyses from chromosome analysis to gene mutation detection, to agreed best practice standards.

## 2.6.2 Learning outcomes

### Learning outcome 1

Demonstrate the ability to perform constitutional chromosome analysis across a range of clinical settings.

### Learning outcome 2

Demonstrate the ability to accurately analyse the results of FISH and array preparations across an appropriate range of clinical settings.

### Learning outcome 3

Understands the advantages and limitations of different commonly used mutation detection techniques and demonstrate the ability to perform a range of these techniques: understand when it is appropriate to utilise a particular methodology.

### Learning outcome 4

Demonstrate the ability to perform gene dosage using MLPA and be able to analyse the results using appropriate software.

### Learning outcome 5

Demonstrate the ability to perform and analyse the results of microsatellite testing, this could include QF-PCR and problems of maternal cell contamination, uniparental disomy studies, microsatellite instability, post Bone Marrow Transplant monitoring or basic linkage analysis.

### Learning outcome 6

Demonstrate an understanding of the principles, application and interpretation of triplet repeats, including the role and application of Southern blotting and methylation analysis.

## 2.6.3 Detailed Content

### Learning Outcome 1

Demonstrate the ability to perform constitutional chromosome analysis across a range of clinical settings.

#### Indicative content and practical skills to be delivered in the workplace

- Chromosome (homologue) identification and compilation of a karyotype.
- Accurate band by band analysis of chromosomes.
- Knowledge of numerical and structural chromosome anomalies and their clinical applications.
- Professional Best Practice Guidelines as applied to constitutional cytogenetics and the role of UKNEQAS.
- Chromosome morphology including polymorphic variants.
- Know, understand and use the International System for Chromosome Nomenclature (ISCN).

## **Learning Outcome 2**

Demonstrate the ability to accurately analyse the results of FISH and array preparations across an appropriate range of clinical settings.

### **Indicative content and practical skills to be delivered in the workplace**

- Knowledge of the range and application of types of available FISH probes.
- Know the range of common clinical applications of FISH in arrays.
- Understand and adhere to Professional Best Practice Guidelines.
- Accurate use of analytical software associated with array platforms in use, including analysis settings and parameters.
- Completion and storage of all audit data including patient records.

## **Learning Outcome 3**

Understands the advantages and limitations of different commonly used mutation detection techniques and demonstrate the ability to perform a range of these techniques: understand when it is appropriate to utilise a particular methodology.

### **Indicative content and practical skills to be delivered in the workplace**

- Principles of mutation detection – advantages and limitations of commonly used techniques to identify novel and known mutations.
- Understand the principles of sequencing chemistries e.g. Sanger sequencing, pyrosequencing, next generation sequencing. Parameters and applications in both manual and automated settings, including limitations.
- Understand the chemistry and explain the methods for mutation scanning e.g. High Resolution Melt analysis (HRM), Denaturing High Performance Liquid Chromatography (DHPLC) and Conformation Sensitive Capillary Electrophoresis (CSCE).
- Perform sequencing reactions – manual and/or robotic.
- Undertake mutation scanning techniques appropriate to the local laboratory repertoire
- Perform techniques to detect known mutations for example Amplification Refractory Mutation Detection System (ARMS), Oligonucleotide Ligation Assay (OLA) and restriction digestion.
- Appropriately use software packages for analysis of results generated.
- Accurately use Human Genome Variation Society (HGVS) mutation nomenclature.

## **Learning Outcome 4**

Demonstrate the ability to perform gene dosage using MLPA, and be able to analyse the results using appropriate software.

### **Indicative content and practical skills to be delivered in the workplace**

- Principles and application of MLPA in a range of clinical settings.
- Demonstrate ability to perform MLPA reactions.

- Apply relevant software to the analysis of the results.

### **Learning Outcome 5**

Demonstrate the ability to perform and analyse the results of microsatellite testing with reference to the quality of the result and an understanding of the clinical context, this could include QF-PCR and problems of maternal cell contamination, uniparental disomy studies, microsatellite instability, post Bone Marrow Transplant monitoring or basic linkage analysis.

#### **Indicative content and practical skills to be delivered in the workplace**

- Microsatellite repeats, their application to copy number detection, in various clinical settings for example aneuploidy detection.
- Gene tracking (linkage analysis) through families and the problems associated with their use.
- Use of appropriate fragment analysis software to determine microsatellite peak size and area when applied to QF-PCR and gene tracking in families.
- How to construct haplotypes on pedigrees.
- Identify low level mixed genotypes e.g. maternal cell contamination, BMT chimerism

### **Learning Outcome 6**

Demonstrate an understanding of the principles, application and interpretation of triplet repeats including the role and application of Southern blotting and methylation analysis.

#### **Indicative content and practical skills to be delivered in the workplace**

- The triplet repeat disorders (e.g. fragile X, Huntington disease, myotonic dystrophy and the spinocerebellar ataxias) and the relevance of mutation type and size to disease presentation and progression.
- Perform fluorescent based PCR detection methods for triplet repeats, relevant to the laboratory. Know the limitations of size detection.
- Use of Southern blotting for the detection of large triplet expansions.
- Process and principles of Southern blotting to include restriction digests, gel electrophoresis, blotting, probe labelling and hybridisation by radioactive and/or non-radioactive methods, stringency washing and signal detection.
- Use of methylation sensitive restriction digests and Southern blotting to detect methylation difference.
- Use of bisulphite modification, methylation-specific PCR, sequencing and MLPA to detect methylation differences.

## **2.7 MODULE 3 Sample Reporting and Interpretation**

This module has 4 learning outcomes. These are shown together to demonstrate the structure of the overall module and then the detailed knowledge and competencies attached to each learning outcome are detailed.

### 2.7.1 Aim

To prepare the trainee to accurately interpret and report the outcome of routine test results.

### 2.7.2 Learning outcomes

#### Learning outcome 1

Demonstrate the ability to validate results from a range of procedures to inform repeat analysis, need for additional investigations, or reporting.

#### Learning outcome 2

Demonstrate the ability to collate all relevant data from analysis software packages and worksheets in readiness for reporting patient results.

#### Learning outcome 3

Demonstrate the ability to draft routine reports for validation, prioritise reports and identify cases for referral to appropriate senior colleague.

#### Learning outcome 4

Recognise and demonstrate the importance of acting within the roles and responsibilities of a HCSP; have a clear understanding of when to seek advice from more senior colleagues.

### 2.7.3 Detailed Content

#### Learning Outcome 1

Demonstrate the ability to validate results from a range of procedures to inform repeat analysis, need for additional investigations, or reporting.

#### Indicative content and practical skills to be delivered in the workplace

- Know and apply the quality parameters which must be met for each test /investigation (including any relevant Best Practice Guidelines).
- Local policies and strategies for stepwise investigations of clinical referrals.

#### Learning Outcome 2

Demonstrate the ability to collate all relevant data from analysis software packages and worksheets in readiness for reporting patient results.

#### Indicative content and practical skills to be delivered in the workplace

- The process and audit trail for identifying that all relevant analysis has been undertaken for a referral.
- The processes for identifying validated results for routine referrals ready for reporting and identifying any outstanding tests requiring completion.
- Collation of data into a format suitable for reporting.

### **Learning Outcome 3**

Demonstrate the ability to draft routine reports for validation, prioritise reports and identify cases for referral to appropriate senior colleague.

#### **Indicative content and practical skills to be delivered in the workplace**

- The process for identifying referrals ready for reporting, including prioritisation of urgent referrals.
- Reasons for upward referral of results for review by senior colleagues.
- Correct selection of reporting template relevant to reason for referral.
- Use of databases to import/upload demographics and/ or results into correct template.
- Checking and authorisation procedures.
- Use of databases for audit of reporting data.

### **Learning Outcome 4**

Recognise and demonstrate the importance of acting within the roles and responsibilities of a HCSP; have a clear understanding of when to seek advice from more senior colleagues.

#### **Indicative Knowledge and Practical skills to be delivered in the workplace**

- Recognises and accepts the responsibilities and role of the Healthcare Science Practitioner in relation to other healthcare professionals.
- Demonstrates increasing ability to prioritise and organise duties in order to optimise the work of the department.

## **2.8 Additional Generic Competencies**

There are a number of generic competencies, which focus on attitudes, behaviours, knowledge and skills that will be delivered throughout the training programme. These are around managerial and leadership skills and health and safety and will be assessed throughout the work based assessment programme.

- Knows how to communicate succinctly and effectively with other professionals as appropriate.
- Demonstrates improving ability to make appropriate decisions in order to optimise the effectiveness of the scientific team resource.
- Understands the structure of the NHS and the management of local healthcare systems in order to be able to participate fully in managing healthcare provision.
- Understands that patient safety depends on the effective and efficient organisation of care, healthcare staff working well together and safe systems not just individual competency and safe practice.
- Recognises the causes of error and to learn from them, realising the importance of honesty and effective apology.

### 3.0 Timetable

The timing of the modules can be quite flexible as individual laboratories may choose to run learning outcomes from different modules together. A suggested timetable is as follows.

<b>Year 1</b>	<b>Module 1</b>	<b>9 months</b>
<b>Year 2</b>	<b>Modules 2 &amp; 3</b>	<b>12 months</b>

### 4.0 Trainee Portfolio, Assessment and Competency Strategy

Further details of the assessment tools and processes can be seen in the accompanying Assessment manual, 'Online assessment and personal development management system' which is available on request from the National School by emailing [Genetics.nwd@westmidlands.nhs.uk](mailto:Genetics.nwd@westmidlands.nhs.uk).

There will be continuous assessment across the 2 year period using a series of validated tools. Trainees will be expected to keep a record of all assessments and competencies in their e-training portfolio. The third section of the portfolio is for the trainees to record any reflective learning and document any additional information.

The competencies are documented in the section 5 of this manual and should be filled in as the trainee progresses through the training programme.

The online assessment tools are based on Direct Observational Skills (DOPS) and Case Based Discussions (CbD).

Case based discussion (CbD) is designed to provide structured teaching and feedback in a particular area of clinical or scientific practice by evaluating decision making and the interpretation and application of evidence. It also enables the discussion of the context, professional, ethical and governance framework of practice, and in all instances, it allows trainees to discuss why they acted as they did. CbDs are used throughout training and should encourage a reflective approach to learning.

Direct observation of practical skills (DOPS) is the observation and evaluation of a procedural/technical or practical skill performed by a trainee in a live environment.

The table below shows the number of DOPS and CbDs within each module. These will be done online and entered into a national database. Each trainee will have an individual password and will be responsible for ensuring these assessments are carried out.

The following indicate the minimum number of formal assessments that should be completed per module. All of these will be in the online portfolio.

Module 1	Module 2	Module 3
2 DOPS Competencies	2 DOPS 1 CBD Competencies	2 DOPS 1 CBD Competencies

Examples of the DOPS and CBD forms are shown at **Appendix 3** and will be available online.

Trainees will be expected to maintain their own portfolio and ensure all assessments are done on time. There will be a national progress review after the first year and a final year assessment, the detail of which will be sent to the trainees during the course.

Trainees will be expected to undertake independent study and a list of suggested educational resources are included in **Appendix 4**.

## 5.0 Competencies

The competencies form the foundation of the training programme, enabling both the trainee and trainer to monitor progress; they are an important part of the e-portfolio and the trainee's record of competence.

It is essential to understand that competency is assessed throughout and is relative to the level of experience at a particular point. Depth of understanding and ability to perform a task will develop as the training period continues. Satisfactory performance is therefore relative. For example, with chromosome analysis the competency will progress from pairing and identification of chromosomes to aneuploidy to large structural rearrangements to small and subtle imbalances. This applies throughout. Your trainer can de-construct individual competences locally to make them time specific. The sign-off for the portfolio signals that the specified level of competence has been achieved. Some competences are therefore 'exit' competences. These are described as such in the recognition that they require longer time and experience to acquire and therefore can't be limited to one specific module or individual learning outcome. Sign off won't be concomitant with the end of the module but will occur some time later.

This manual provides some examples of evidence or application required in order to demonstrate competence. It is not definitive. Trainees are expected to utilise different tools, resources and media within the local laboratory to demonstrate evidence of each competence.

Competencies are transferable across learning outcomes and do not need to be undertaken twice where they are repeated.

Where verbs such as recognise, demonstrate, know and similar are used in the table below it means to do these accurately and consistently.

## 5.1 GENERIC COMPETENCIES

These apply to all (or most modules).

Code	Competency	Reviewer	Date	Comments
G1.1	Use and maintain laboratory equipment specific to the section safely and appropriately.			
G1.2	Describe and undertake accurate stock control and quality control measures for the consumables specific to the section in which working.			
G1.3	Use the laboratory database(s) effectively.			
G1.4	Recognise the hazards associated with biological samples and handle them appropriately in order to minimise risk.			
G1.5	Recognise high risk samples and follow local policies for high risk samples.			
G1.6	Prepare reagents and media accurately, adhering to protocols.			
G1.7	Be able to process samples in a timely fashion based on clinical need.			
G1.8	Be able to store samples appropriately at all stages during and after the investigation.			

## 5.2 Examples of Application

To evidence competencies trainees must be able to provide examples of how the knowledge or skill has been applied in a practical setting. The list below provides some examples of application but trainees are expected to utilise a variety of different tools, resources and media within the local laboratory to demonstrate evidence of each competency;

- Aware of relevant Standard Operating Procedures (SOPS) and adheres to them.
- Direct assessment and account of assessor.
- Provide advice – verbal, written or by telephone to a response or query.
- Management of referrals in an appropriate manner i.e., brought to the attention of an appropriate member of staff.
- Management of samples and use of local systems to record.
- Discussion between trainee and trainer.
- Accurate management of patient data and sample information.
- Peer observation.

- Examples of problem solving/trouble shooting.

### 5.3 Exit Competencies

Exit competencies are recognised as competencies, which require longer time and experience to acquire beyond the specific module in which they appear in the programme. Trainees will not be required to demonstrate competence until such point that the trainer considers the trainee to have achieved the level and skill required to be considered competent. Sign off won't be concomitant with the end of the module but will occur some time later.

Exit competencies are demonstrated here for clarity. These competencies will also appear within the module to which they relate in the programme.

Code	Competency	Reviewer	Date	Comments
E1.1/ M2.1. 2	Analyse G-banded metaphase preparations accurately and within an acceptable timescale by completing an analysis test			
E1.2/ M3.1. 1	To describe and apply the correct quality parameters (e.g. control data, peak height and area, minimum fluorescence etc) to determine the validity of the range of test results.			
E1.3/ M3.1. 1	Describe the local policy for repeat analysis and/or application of follow-up tests to verify results.			
E1.4/ M3.1. 3	Identify validated results, which are ready for reporting, demonstrating application of policies for testing scenarios (e.g. predictive or diagnostic tests).			
E1.5/ M3.2. 1	Undertake a full audit trail to identify all data relevant to reporting of a patient result.			
E1.6/ M3.2. 2	Recognise sufficiency of tests for routine reporting and when to seek advice from a senior colleague.			

### 5.4 Module 1 Sample Receipt and Preparation

#### 5.4.1 Learning Outcome 1

Demonstrate understanding and skills in the principles of safe receipt and handling of samples including data entry and storage.

Code	Competency	Reviewer	Date	Comments
M1.1.1	Recognise the hazards associated with biological samples and handle them appropriately in order to minimise risk.			
M1.1.2	Recognise high risk samples and alert senior staff.			
M1.1.3	Describe containers that are suitable for samples, given the nature of the test requested and transport requirements.			
M1.1.4	Demonstrate awareness of the local policies for the acceptance and processing of samples.			
M1.1.5	Recognise the condition of samples and their suitability for processing.			
M1.1.6	Know when to refer decisions for processing suboptimal samples to senior colleagues.			
M1.1.7	Demonstrate knowledge of different types of sample tubes, and their appropriateness.			
M1.1.8	Describe and follow local policies regarding patient/sample data entry.			
M1.1.9	State and adhere to local policy regarding patient confidentiality.			
M1.1.10	Describe how manual and electronic records are stored within the laboratory and apply correct procedures to access them.			
M1.1.11	Describe and implement procedures for obtaining essential missing information (excluding clinical information).			
M1.1.12	Use the laboratory database(s) effectively to make general enquiries.			
M1.1.13	Adhere to local procedures for waste disposal, recycling, decontamination and spillages.			
M1.1.14	Describe and perform correct procedures for storage of samples prior to processing.			
M1.1.15	Understand the policies and procedures for the onward referral of samples and apply accurately and in			

	a timely manner.			
M1.1.16	Demonstrate awareness of the need for patient consent and adhere to local policy.			

#### 5.4.2 Learning Outcome 2

Demonstrate understanding and skill in cell culturing, harvesting, slide making and staining techniques for chromosome and genetic analysis (including both short and long term culture)

Code	Competency	Reviewer	Date	Comments
M1.2.1	Describe and undertake preparation of culture media and reagents.			
M1.2.2	Recognise high risk samples and follow local policies for high risk samples for their handling at each stage.			
M1.2.3	Recognise the condition of samples, their suitability for culture and the appropriate variations, which may need to be employed if the sample is substandard.			
M1.2.4	Describe and use correct aseptic technique.			
M1.2.5	Describe and perform setting up of short and long term cultures to defined laboratory standards.			
M1.2.6	Describe and perform assessment of ongoing long term cultures (where appropriate).			
M1.2.7	Describe and perform harvesting of short and long term cultures to defined laboratory standards.			
M1.2.8	Describe and perform slide making for genetic analysis from cultures to defined laboratory standards to include troubleshooting			
M1.2.9	Describe and perform G-banding of chromosome preparations to defined laboratory standards.			
M1.2.10	Describe and perform correct procedures for storage of samples awaiting processing.			
M1.2.11	Describe and perform correct disposal of all waste material arising from cell culture.			

M1.2.12	Describe and perform correct storage of fixed cell suspensions following cell culture.			
M1.2.13	Describe and perform correct long term storage of slides.			

### 5.4.3 Learning Outcome 3

Demonstrate understanding and skill in DNA and RNA extraction from a range of patient samples types. Evaluate the quality and quantity required for subsequent analyses.

Code	Competency	Reviewer	Date	Comments
M1.3.1	Use appropriate methods to control and eliminate microbial or chemical contamination from working environment.			
M1.3.2	Practice measures that prevent cross-contamination between samples.			
M1.3.3	Identify correct extraction method for referral type.			
M1.3.4	Isolate DNA/RNA expediently, with consideration to specimen type and test required using the appropriate method.			
M1.3.5	Reconstitute precipitated DNA/RNA using appropriate buffer and dilute according to local policy for sample type and referral reason.			
M1.3.6	Determine quality and quantity of DNA/RNA using spectrophotometric methods.			
M1.3.7	Use gel electrophoresis to estimate concentration and determine quality and integrity of DNA/RNA.			
M1.3.8	Describe stock control and quality control measures for consumables specific to the section.			
M1.3.9	Describe (and undertake) preparation of extraction reagents.			
M1.3.10	Maintain database and paper records to document outcome of extraction procedures.			
M1.3.11	Describe and perform procedures for storage of samples awaiting processing.			
M1.3.12	Describe and perform correct disposal of waste material arising			

	from DNA/RNA extraction. Include awareness of Human Tissue Act (HTA).			
M1.3.13	Describe and perform procedure for correct long term storage of DNA/RNA.			

#### 5.4.4 Learning Outcome 4

Understand the principles of and process samples for array analysis using a variety of platforms such as SNP, OLIGO and resequencing arrays, depending on local availability

Code	Competency	Reviewer	Date	Comments
M1.4.1	Use and maintain array scanner following appropriate protocols.			
M1.4.2	Use and maintain other laboratory equipment specific to the section.			
M1.4.3	Describe and correctly perform preparation of reagents used in microarray testing.			
M1.4.4	Accurately measure and assess the quantity and quality of DNA relevant to the task to be undertaken.			
M1.4.5	Describe and perform labelling reaction for microarray to defined laboratory standards.			
M1.4.6	Describe and perform the range of practical steps for the processing of samples for microarray testing to defined laboratory standards, either manually or automated as in use in the training laboratory.			
M1.4.7	Describe and perform loading of array and appropriate associated data onto scanner and other associated databases.			
M1.4.8	Describe and perform retrieval of results data from array scanner (if appropriate).			

#### 5.4.5 Learning Outcome 5

Prepare and successfully process an appropriate range of in situ hybridisation probes (FISH), for example, chromosome enumeration probes, locus specific probes and whole chromosome paints.

Code	Competency	Reviewer	Date	Comments
M1.5.1	Recognise sub-optimal FISH preparations, understand the reasons for this and undertake remedial action.			
M1.5.2	Describe and perform FISH hybridisation to defined laboratory standards on constitutional preparations.			

#### 5.4.6 Learning Outcome 6

Demonstrate the ability to perform a range of PCR techniques manually and using automated technology, including sample preparation and validation of the outcome of the reaction.

Code	Competency	Reviewer	Date	Comments
M1.6.1	Explain the need for and use pre and post PCR areas of the lab effectively.			
M1.6.2	Understand the suitable conditions for storage and working with fluorescently – labelled oligonucleotides.			
M1.6.3	Describe and use standard procedures for preparing primer mixes for PCR, including recording the appropriate details of primers where needed (batch records).			
M1.6.4	Describe and perform setting up of PCR to defined laboratory standards, both manual and automated.			
M1.6.5	Demonstrate awareness of factors affecting PCR quality.			
M1.6.6	Demonstrate awareness of sensitivity to contamination and describe measures employed to minimise risk.			
M1.6.7	Describe how the procedures may be adjusted to correct for poor PCR quality (troubleshooting).			
M1.6.8	Describe and perform loading of PCR samples onto thermal cycler.			
M1.6.9	Describe and use basic programming functions of thermal			

	cycler.			
M1.6.10	Understand principles of and perform agarose gel electrophoresis and size separation, explain effect of voltage and current buffers, use appropriate sizing markers.			

## 5.5 Module 2 Sample Analysis – From Chromosomes to Genes

### 5.5.1 Learning Outcome 1

Demonstrate the ability to perform constitutional chromosome analysis across a range of clinical settings.

Code	Competency	Reviewer	Date	Comments
M2.1.1	Recognise chromosomes of the appropriate quality level for the reason for referral.			
E1.1/ M2.1.2	Analyse G-banded metaphase preparations accurately and within an acceptable timescale by completing an analysis test			
M2.1.3	Understand and employ the local policy for constitutional chromosome analysis.			
M2.1.4	Demonstrate a working knowledge of brightfield microscopy and image analysis systems.			
M2.1.5	Demonstrate awareness and use of ACC Best Practice Guidelines relevant to constitutional chromosome analysis.			
M2.1.6	Use current ISCN to describe normal and abnormal constitutional karyotypes.			
M2.1.7	Recognise and describe the range of polymorphic chromosome variation.			
M2.1.8	Demonstrate knowledge of commonly encountered chromosome findings, including mosaicism, marker chromosomes and fragile sites.			
M2.1.9	Recognise and describe situations when additional banding techniques or FISH would provide extra beneficial information.			

### 5.5.2 Learning Outcome 2

Demonstrate the ability to accurately analyse the results of FISH and array preparations across an appropriate range of clinical settings.

Code	Competency	Reviewer	Date	Comments
M2.2.1	Describe the categories of FISH probes commonly used in genetic diagnosis. Understand the advantages and limitations of each.			
M2.2.2	Describe the range of microarray platforms commonly used in genetic diagnosis for copy number variation. Understand the advantages and limitations of each.			
M2.2.3	Analyse FISH interphase and metaphase preparations in an accurate and timely manner, in accordance with national best practice guidelines.			Commercially available probes and paints only
M2.2.4	Analyse microarray results in an accurate and timely manner in accordance with national best practice guidelines and local policies.			

### 5.5.3 Learning Outcome 3

Understands the advantages and limitations of different commonly used mutation detection techniques and demonstrate the ability to perform a range of these techniques: understand when it is appropriate to utilise a particular methodology.

Code	Competency	Reviewer	Date	Comments
M2.3.1	Describe the range of commonly used mutation detection techniques employed in diagnostic laboratories			
M2.3.2	Perform ARMS or OLA to detect known mutations			
M2.3.3	Understand and, if available locally, be able to perform scanning techniques, such as HRM or dHPLC to detect unknown mutations.			
M2.3.4	Perform sequence reaction set up either manually or using automated platforms.			

M2.3.5	Be able to analyse data generated using different detection methods and platforms using appropriate software, for example Mutation Surveyor sequence analysis and Alamut software use.			
M2.3.6	To understand and apply correct HGVS mutation nomenclature to detected sequence variants.			
M2.3.7	Be able to recognise good quality and poor quality data from the analysis process using defined quality parameters and perform necessary trouble-shooting.			
M2.3.8	Be able to correctly record the outcome of the analysis.			
M2.3.9	Trouble-shoot and perform routine preventative maintenance on different platforms, relevant to the laboratory			
M2.3.10	Know and understand health and safety issues relating to each technique/platform.			
M2.3.11/ G1.7	Be able to process samples in a timely fashion based on clinical need.			
M2.3.12/ G1.8	Be able to store samples appropriately at all stages of the investigation and thereafter			
M2.3.13	Be familiar with participation in EQA schemes appropriate to the platform/technique.			
M2.3.14	Understand and perform housekeeping roles relating to working area/platform.			
M2.3.15	Upkeep of batch records as necessary to maintain Quality Control.			
M2.3.16	Understand and perform stock control of reagents etc as necessary.			

#### 5.5.4 Learning Outcome 4

Demonstrate the ability to perform gene dosage by MLPA, and be able to analyse the results using appropriate software.

Code	Competency	Reviewer	Date	Comments
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M2.4.1	Describe and understand the principles of MLPA to detect gene dosage.			
M2.4.2	Use and perform routine maintenance of laboratory equipment relevant to MLPA.			
M2.4.3	Set up MLPA reaction to detect dosage differences in a variety of clinical referrals.			
M2.4.4	Be able to apply correct software to analyse MLPA data.			
M2.4.5	Be able to recognise good quality and poor quality data from the analysis process, using defined quality parameters and perform necessary trouble-shooting.			
M2.4.6	Be able to correctly record the outcome of the analysis.			
M2.4.7	Understand health and safety issues relating to technique/platform.			
M2.4.8/ G1.7	Be able to process samples in a timely fashion based on clinical need.			
M2.4.9/ G1.8	Be able to store samples appropriately at all stages of the investigation and thereafter.			
M2.4.10	Undertake housekeeping roles relating to working area/platform.			
M2.4.11	Maintain batch records as necessary to maintain Quality Control.			
M2.4.12	Know, understand and perform the required QA of platform/technique.			
M2.4.13	Understand and perform stock control of reagents etc as necessary.			

### 5.5.5 Learning Outcome 5

Demonstrate the ability to perform and analyse the results of microsatellite testing with reference to the quality of the result and an understanding of the clinical context, this could include QF-PCR and problems of maternal cell contamination, uniparental disomy studies, microsatellite instability, post Bone Marrow Transplant monitoring or basic linkage analysis

Code	Competency	Reviewer	Date	Comments
M2.5.1	Be able to select appropriate			

	microsatellite markers or kits for the analysis of referrals for QF-PCR, UPD, maternal cell contamination or linkage analysis.			
M2.5.2	Be able to define the nature of microsatellite markers and how they can be used to track inheritance of alleles within a pedigree.			
M2.5.3	To describe the referral reasons and/or clinical context of referrals for QF PCR, MRD, UPD, maternal cell contamination and linkage analysis.			
M2.5.4	To describe the quality parameters relevant to the application of the test (i.e. the sensitivity of the test in the context of the clinical question being asked).			
M2.5.5	To perform the full range of tests using the appropriate platform.			
M2.5.6	To accurately analyse and record the results of the analysis using the appropriate tools and software.			
M2.5.7	Maintain batch records as necessary to maintain Quality Control.			
M2.5.8	Know, understand and perform the required QA of platform/technique.			
M2.5.9	Understand and perform stock control of reagents etc as necessary.			
M2.5.10	Know and understand other techniques/platforms not used in base laboratory.			
M2.5.11	Understand health and safety issues relating to technique/platform.			
M2.5.12/ G1.7	Be able to process samples in a timely fashion based on clinical need.			
M2.5.13/ G1.8	Be able to store samples appropriately at all stages of the investigation and thereafter			
M2.5.14	Undertake housekeeping roles relating to working area/platform.			

### 5.5.6 Learning Outcome 6

Demonstrate an understanding of the principles, application and interpretation of triplet repeats, role and application of Southern blotting and methylation analysis.

Code	Competency	Reviewer	Date	Comments
M2.6.1	To briefly describe the molecular basis of Fragile X syndrome, Myotonic dystrophy and Prader Willi and Angelman's syndromes in relation to the common referral reasons for each condition (for example diagnostic testing for males and females in Fragile X, carrier testing for Fragile X, identification of normal transmitting males in Fragile X, prenatal diagnosis).			
M2.6.2	To describe the principles and practice of restriction digest, electrophoresis and Southern blotting as applied to the analysis of triplet repeats and to look at methylation differences.			
M2.6.3	To understand the limitations and common problems encountered in Southern blotting, which might affect interpretation.			
M2.6.4	To demonstrate the ability to correctly assign alleles for the common clinical scenarios for referrals for Fragile X.			
M2.6.5	To demonstrate the ability to identify results of suboptimal standard and provide suggestion for improvement of quality.			

## 5.6 Module 3 Sample Reporting and Interpretation

### 5.6.1 Learning Outcome 1

Demonstrate the ability to validate results from a range of procedures to inform repeat analysis, need for additional investigations, or reporting.

Code	Competency	Reviewer	Date	Comments
M3.1.1/ E1.2	To describe and apply the correct quality parameters (e.g. control data, peak height and area, minimum fluorescence etc) to determine the validity of the range of test results.			This is an exit competence

M3.1.2/ E1.3	Describe the local policy for repeat analysis and/or application of follow-up tests to verify results.			This is an exit competence
M3.1.3/ E1.4	Identify validated results, which are ready for reporting, demonstrating application of policies for testing scenarios (e.g. predictive or diagnostic tests).			This is an exit competence

### 5.6.2 Learning Outcome 2

Demonstrate the ability to collate all relevant data from analysis software packages and/or worksheets in readiness for reporting patient results.

Code	Competency	Reviewer	Date	Comments
M3.2.1/ E1.5	Undertake a full audit trail to identify all data relevant to reporting of a patient result.			This is an exit competence
M3.2.2/ E1.6	Recognise sufficiency of tests for routine reporting and when to seek advice from a senior colleague.			This is an exit competence
M3.2.3/ E1.7	Collate validated results in correct format for reporting.			

### 5.6.3 Learning Outcome 3

Demonstrate the ability to draft routine reports for validation, prioritise reports and identify cases for referral to appropriate senior colleague.

Code	Competency	Reviewer	Date	Comments
M3.3.1	Demonstrate a basic understanding of the clinical features of common genetic disorders relevant to the reports issued.			
M3.3.2	Demonstrate an understanding of the main reasons for routine referrals across a range of tests.			
M3.3.3	Describe local policy for reporting results of normal and abnormal chromosome analysis on constitutional samples.			
M3.3.4	Ability to draft routine reports on normal and abnormal chromosome analysis on constitutional samples.			

M3.3.5	Select the correct reporting template relevant to reason for referral for results of molecular investigations.			
M3.3.6	Understand the clinical impact of selecting an incorrect template.			
M3.3.7	Apply local procedures for the prioritisation of reports.			
M3.3.8	Demonstrate the ability to identify referrals requiring input from senior colleagues.			

#### 5.6.4 Learning Outcome 4

Recognise and demonstrate the importance of acting within the roles and responsibilities of a HCSP; have a clear understanding of when to seek advice from more senior colleagues.

Code	Competency	Reviewer	Date	Comments
M3.4.1	Understands the structure, and the range of roles and responsibilities within the department.			
M3.4.2	Understands own role, responsibility and level of experience.			
M3.4.3	Consistently identify problems /issues requiring referral to a more senior colleague.			

#### 5.7 Additional Generic Competencies

There are a number of generic competencies, which focus on attitudes, behaviours, knowledge and skills, which will be delivered throughout the programme tested. For the PTP programme these are around Managerial and Leadership skills and health and safety. These will be assessed throughout the work based assessment programme and can be demonstrated in the DOPs and CbDs.

Code	Competency	Reviewer	Date	Comments
G2.1	Recognise and accept the responsibilities and role of the Healthcare Practitioner in relation to other healthcare professionals.			
G2.2	Communicate succinctly and effectively with other professionals as appropriate.			
G2.3	Demonstrate increasing ability to prioritise and organise duties in order to optimise the work of the department.			

G2.4	Demonstrate improving ability to make appropriate decisions in order to optimise the effectiveness of the scientific team resource			
G2.5	Understands the structure of the NHS and the management of local healthcare systems in order to be able to participate fully in managing healthcare provision.			
G2.6	Understand that patient safety depends on the effective and efficient organisation of care, healthcare staff working well together and safe systems not just individual competency and safe practice.			
G2.7	Recognise the causes of error and to learn from them, realising the importance of honesty and effective apology.			
G2.8	Recognise the desirability of monitoring performance, learning from mistakes and adopting a no blame culture in order to ensure high standards of care and optimise patient safety.			

## The Domains of Good Scientific Practice

### 1.1 Professional Practice

- 1.1.1 Make the patient your first concern
- 1.1.2 Exercise professional duty of care
- 1.1.3 Work within the agreed scope of practice for lawful, safe and effective healthcare science
- 1.1.4 Keep your professional, scientific, technical knowledge and skills up to date
- 1.1.5 Engage fully in evidence based practice
- 1.1.6 Draw on appropriate skills and knowledge in order to make professional judgements
- 1.1.7 Work within the limits of your personal competence
- 1.1.8 Be open and honest and act with integrity
- 1.1.9 Act without delay if you have good reason to believe that you or a colleague may be putting people at risk
- 1.1.10 Never discriminate unfairly against patients or colleagues
- 1.1.11 Maintain your fitness to practice
- 1.1.12 Treat patients as individuals, respect their dignity and confidentiality and uphold the rights, values and autonomy of every service user, including their role in the diagnostic and therapeutic process and in maintaining health and well-being.
- 1.1.13 Respond constructively to the outcome of audit, appraisals and performance reviews, undertaking further training where necessary

### 1.2 Scientific

- 1.2.1 Develop investigative strategies/procedures/processes that take account of relevant clinical and other sources of information
- 1.2.2 Provide scientific advice to ensure the safe and effective delivery of services
- 1.2.3 Undertake scientific investigations using qualitative and quantitative methods to aid the screening, diagnosis, prognosis, monitoring and/or treatment of health and disorders appropriate to the discipline
- 1.2.4 Investigate and monitor disease processes and normal states
- 1.2.5 Use and display outcomes from statistical packages that are appropriate to scope of practice
- 1.2.6 Critically evaluate data, draw conclusions from it, formulate actions and recommend further investigations where appropriate

### 1.3 Clinical

- 1.3.1 Ensure that you and the staff you supervise understand the need for and obtain relevant consent before undertaking any investigation, examination, provision of treatment, or involvement of patients in teaching or research
- 1.3.2 Ensure that you and the staff you supervise maintain confidentiality of patient information and records in line with published guidance
- 1.3.3 Ensure that you and your staff understand the wider clinical consequences of decisions made on your actions or advice
- 1.3.4 Demonstrate expertise in the wider clinical situation that applies to patients who present in your discipline

- 1.3.5 Maintain up to date knowledge of the clinical evidence base that underpins the services that you provide and/or supervise and ensure that these services are in line with the best clinical evidence
- 1.3.6 Plan and determine the range of clinical/scientific investigations or products required to meet diagnostic, therapeutic, rehabilitative or treatment needs of patients, taking account of the complete clinical picture
- 1.3.7 Plan and agree investigative strategies and clinical protocols for the optimal diagnosis, monitoring and therapy of patients with a range of disorders
- 1.3.8 Ensure that detailed clinical assessments are undertaken and recorded using appropriate techniques and equipment and that the outcomes of these investigations are reviewed regularly with users of the service
- 1.3.9 Ensure the provision of expert interpretation of complex and or specialist data across your discipline in the context of clinical questions posed
- 1.3.10 Undertake and record a detailed clinical assessment using appropriate techniques and equipment
- 1.3.11 Provide specialised clinical investigation and/or analysis appropriate to your discipline
- 1.3.12 Provide interpretation of complex and/or specialist data in the context of the clinical question posed
- 1.3.13 Provide clinical advice based on results obtained, including a diagnostic or therapeutic opinion for further action to be taken by the individual directly responsible for the care of the patient
- 1.3.14 Provide expert clinical advice to stakeholders in order to optimise the efficiency and effectiveness of clinical investigation of individuals and groups of patients
- 1.3.15 Prioritise the delivery of investigations, services or treatment based on clinical need of patients
- 1.3.16 Represent your discipline in multidisciplinary clinical meetings to discuss patient outcomes and the appropriateness of services provided
- 1.3.17 Ensure that regular and systematic clinical audit is undertaken and be responsible for modifying services based on audit findings.

#### **1.4 Technical**

- 1.4.1 Provide technical advice to ensure the safe and effective delivery of services
- 1.4.2 Plan, take part in and act on the outcome of regular and systematic audit
- 1.4.3 Work within the principles and practice of instruments, equipment and methodology used in the relevant scope of practice
- 1.4.4 Demonstrate practical skills in the essentials of measurement, data generation and analysis
- 1.4.5 Assess and evaluate new technologies prior to routine use
- 1.4.6 Use tables and graphs in order to analyse experimental data
- 1.4.7 Identify and manage sources of risk in the workplace, including specimens, raw materials, clinical and special waste, equipment, radiation risks and electrical risks
- 1.4.8 Apply principles of good practice in health and safety to all aspects of the workplace
- 1.4.9 Apply correct principles and applications of disinfectants, methods for sterilisation, decontamination and for dealing with waste and spillages correctly.
- 1.4.10 Demonstrate appropriate level of skill in the use of information technology appropriate to practice

## **1.5 Investigation and reporting**

- 1.5.1 Plan and conduct scientific, technical, diagnostic, monitoring, treatment and therapeutic procedures with professional skill and ensuring the safety of patients, the public and staff
- 1.5.2 Perform investigations and procedures/design products to assist with the management, diagnosis, treatment, rehabilitation or planning in relation to the range of patient conditions/equipment within a specialist scope of practice
- 1.5.3 Monitor and report on progress of patient conditions/use of technology and the need for further interventions.
- 1.5.4 Interpret and report on a range of investigations or procedures associated with the management, of patient conditions/equipment

## **1.6 Quality**

- 1.6.1 Set, apply and maintain and apply quality standards, control and assurance techniques for interventions across all clinical, scientific and technological activities
- 1.6.2 Make judgements on the effectiveness of procedures, processes
- 1.6.3 Participate in quality assurance programmes
- 1.6.4 Maintain an effective audit trail and work towards continuous improvement

## **1.7 Working with colleagues**

- 1.7.1 Work with other professionals, support staff, service users, carers and relatives in the ways that best serve patients' interests
- 1.7.2 Work effectively as a member of a multi-disciplinary team
- 1.7.3 Consult and take advice from colleagues where appropriate
- 1.7.4 Be readily accessible when you are on duty
- 1.7.5 Respect the skills and contributions of your colleagues
- 1.7.6 Participate in regular reviews of team performance and take steps to remedy any deficiencies

## **1.8 Research and development**

- 1.8.1 Search and critically appraise scientific literature and other sources of information
- 1.8.2 Engage in evidence-based practice and participate in audit procedures
- 1.8.3 Apply a range of research methodologies and initiate and participate in collaborative research
- 1.8.4 Manage research and development according within a governance framework
- 1.8.5 Evaluate, validate and verify new scientific, technical, diagnostic, monitoring, treatment and therapeutic procedures
- 1.8.6 Evaluate research and other evidence to inform own practice
- 1.8.7 Interpret data in the prevailing clinical context
- 1.8.8 Perform experimental work, produce and present results
- 1.8.9 Present data and research findings to peers in appropriate forms

## **1.9 Probity**

- 1.9.1 Make sure that your conduct at all times justifies the trust of patients and colleagues and maintains the public's trust in the scientific profession
- 1.9.2 Inform your statutory authority without delay if, at any time, you have accepted a caution, been charged with or found guilty of a criminal offence, or if any finding has been made against you as a result of fitness to practice procedures, or if you are

suspended from a scientific post, or if you have any restrictions placed on your scientific, clinical or technical practice

- 1.9.3 Be honest and trustworthy when writing reports or signing documents
- 1.9.4 Be honest about your qualifications, experience, and position in the scientific community
- 1.9.5 Take all reasonable steps to verify information in reports and documents, including research
- 1.9.6 Be honest in written and verbal information provided to any formal enquiry or litigation, including that relating to the limits of your scientific knowledge and experience.
- 1.9.7 Work within the HPC Standards of Conduct, Performance and Ethics

### **1.10 Leadership**

- 1.10.1 Maintain responsibility when delegating healthcare activities and provide support as needed
- 1.10.2 Respect the skills and contributions of your colleagues
- 1.10.3 Protect patients from risk or harm presented by a colleague's conduct, performance or health
- 1.10.4 Treat your colleagues fairly and with respect
- 1.10.5 Make suitable arrangements to ensure that roles and responsibilities are covered when you are absent, including handover at sufficient level of detail to competent colleagues
- 1.10.6 Ensure that patients and colleagues understand the role and responsibilities of each member of the team
- 1.10.7 Ensure that systems are in place through which colleagues can raise concerns
- 1.10.8 Ensure regular reviews of team performance and take steps to remedy any deficiencies
- 1.10.9 Refer patients only to professional staff who can be accountable to a statutory body

### **1.11 Training and developing others**

- 1.11.1 Support colleagues who have difficulties with performance, conduct or health
- 1.11.2 Share information with colleagues to protect patient safety
- 1.11.3 Provide work-based development for colleagues to enhance/improve skills and knowledge
- 1.11.4 Identify and take appropriate action to meet the development needs of those for whom you have management, supervision or training responsibilities

## Role descriptor template for Healthcare Science Practitioner (HCSP)

A Healthcare Science Practitioner (HCSP) will have the necessary expertise in applied scientific techniques within a discipline or related disciplines and will work in a range of healthcare settings:

With a defined role in the delivery and reporting of quality assured tests, investigations and interventions on patients, samples or equipment;

In a number of disciplines, HCSP will provide therapeutic interventions, some of which may be specialist

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### ***General Scope in Genetics***

- HCSPs should work within protocol driven activity.
- A HCSP in genetics will have technical expertise within genetics and will work in a range of healthcare settings related to genetics. HCSPs are likely to work under indirect supervision and be flexible to meet the needs of the service.
- Requires a good knowledge and understanding of relevant scientific and technical principles and practice

### **Scientific and Technical**

- Performing a range of testing including analysis (but where the outcomes/significance are interpreted and reported by healthcare scientists) in a range/combination of:
  - Molecular testing – for example gene sequencing, fragment analysis
  - Gene chips – for example resequencing
  - Molecular cytogenetics – for example arrays, FISH
  - Chromosome analysis
  - Molecular pathology
  - Oncology
  - Utilisation of genomic databases/bioinformatics
- Is responsible for technical and analytical support and will be expected to perform a range of regular duties as well as occasional specific requests for specialised procedures in accordance with defined protocols.
- Perform highly accurate and skilled analysis on a range of genetic material e.g. DNA, RNA and chromosomes derived from a wide range of pathological samples
- Preparation of a wide range of samples using the most up to date technologies appropriate for patient care.
- Perform and validate genetic analysis of samples looking for genetic changes including the use and maintenance of a wide range of technology

- Contribute to the development of protocols in the appropriate technical and scientific areas
- Identifying routine equipment requirements and contribute to the evaluation and commissioning of equipment

### **Patient contact and care**

Requires a basic working knowledge and understanding of clinical situations relevant to specific areas of work, to enable practitioners to:

- Collect clinical samples from patients as appropriate within their care pathway
- Perform point of care testing for patients in a variety of settings
- Compile routine and standard reports of tests for senior scientific staff using clearly defined protocols

### **Administration and Leadership**

- Supervision of small teams on protocol-driven activities
- Stock management of laboratory consumables
- Routine maintenance and quality assurance of technical and scientific equipment
- Day to day organisation of and significant contribution to delivery of analytic service

### **Communication and working with others**

- Communicate appropriately with patients, colleagues and other healthcare professionals and external suppliers
- Liaise with scientists and other healthcare professionals on appropriateness of tests, results and further tests that may be necessary

### **Education and training**

- Technical training of others within your own areas of practice
- Undertaking assessment as appropriate of staff in training

### **Research and development**

- Collection and analysis of samples and data
- Contribute to R&D within the department, including scientific method development and validation
- Contribute to local innovation and technology evaluation

### **Clinical Governance**

- Maintain standards for laboratory health and safety procedures
- Comply with quality and governance procedures within the department including risk
- Maintain high standards of professional and personal conduct
- Ensure that patient safety and experience and effectiveness of service is maximised

- Perform, analyse and report on relevant clinical audits
- HCSPs are responsible for their own work, have a significant role in ensuring the accuracy and quality of the work in their laboratory and work in a range of health care settings

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**Direct Observation of Practical/Procedural Skills for Genetics PTP**

Trainee identification data
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Procedure:	<b>Any procedure carried out by HCSP</b>
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Clinical context:	Sample Receipt & Preparation	Sample Analysis	Sample Reporting & Interpretation
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Assessor's Name:	
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Assessor's position:	
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Difficulty of the procedure:	Low	Average	High
Number of times procedure performed by trainee:	1-4	5-9	>10

Please grade the following areas using the scale below	Below expectations	Borderline	Meets expectations	Above expectations	Unable to comment <sup>1</sup>
1. Understands scientific principles of procedure including basic biology underpinning it					
2. Has read, understands and follows the appropriate SOP's, risk and COSHH assessments, and any other relevant H&S documentation					
3. Understands and applies the appropriate internal and external quality control associated with the procedure					
4. Understands the risks associated with items of equipment and uses them appropriately					
5. Accurately completes associated documentation					
6. Output meets accepted laboratory/professional standards					

7. Carries out the procedure within appropriate time frame					
8. Is aware of the limitations of the test					
9. Demonstrates awareness of the limits of responsibility and when to seek advice					
10. Professionalism					

<sup>1</sup> Unable to comment. Please mark this if you have not observed the behaviour

<b>FEEDBACK AND DOCUMENTATION OF LEARNING NEEDS</b>	<b>AGREED ACTION</b>

<b>Outcome</b>	<b>Satisfactory</b> <b>Unsatisfactory</b>	<b>Date of assessment</b>		<b>Time taken for assessment:</b>	
<b>Signature of Assessor</b>		<b>Signature of Trainee</b>		<b>Time taken for feedback:</b>	

## Case Based Discussion Template for Genetics PTP:

### Trainee identification data

**Brief description of output and focus of scenario discussed:**

<b>Module:</b>	<b>Sample Receipt &amp; Preparation</b>	<b>Sample Analysis</b>	<b>Sample Reporting &amp; Interpretation</b>
<b>Complexity of the scenario:</b>	<b>Low</b>	<b>Average</b>	<b>High</b>

**Assessor's Name:**

**Assessor's position:**

Please grade the following areas using the scale below	Below expectations	Borderline	Meets expectations	Above expectations	Unable to comment <sup>1</sup>
1. Understands clinical and/or scientific principles relevant to scenario					
2. Can discuss relevant health and safety issues					
3. Can discuss the procedures used to obtain the results					
4. Can discuss the quality control procedures to ensure the result is accurate					
5. Demonstrates a knowledge of relevant 'Best Practice' guidelines and other policies relevant to the scenario					
6. Can discuss the significance of routine patient results with reference to the reason for referral					
7. Is aware of, and can use as required, appropriate resources to aid in the interpretation of results					
8. Is aware of importance of audit trail and can complete audit trail accurately					

<b>9. Demonstrates awareness of the limits of responsibility and when to seek advice</b>					
<b>10. Professionalism</b>					

<sup>1</sup> Unable to comment. Please mark this if you have not observed the behaviour

<b>FEEDBACK AND DOCUMENTATION OF LEARNING NEEDS</b>	<b>AGREED ACTION</b>

<b>Outcome</b>	<b>Satisfactory Unsatisfactory</b>	<b>Date of assessment</b>		<b>Time taken for assessment:</b>	
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<b>Signature of Assessor</b>		<b>Signature of Trainee</b>		<b>Time taken for feedback:</b>	
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**Educational Resources****GENETICS AND CELL BIOLOGY**

Molecular Biology of the Gene (5th edition), Watson, Baker, Bell, Gann and Losick (2004) Benjamin Cummings Publishing Co. Inc.

Genes VIII, Lewin (2004) John Wiley

Molecular Cell Biology (5<sup>th</sup> edition), Lodish, ed. (2003) Freeman

**CLINICAL GENETICS**

ABC of Clinical Genetics (3<sup>rd</sup> edition) Kingston (2002) BMJ Books

Medical Genetics. Young (2005), Oxford Medical Publications

Principles and Practice of Medical Genetics (4<sup>th</sup> edition) Emery and Rimoin, Eds. (2002) Churchill Livingstone

Practical Genetic Counselling (6<sup>th</sup> edition), Harper (2004) Arnold publications

Mendelian Inheritance in Man: Catalogues of Autosomal Dominant, Autosomal Recessive, and X-linked Phenotypes. McKusick (1998) Johns Hopkins University Press  
*(This book provides a useful adjunct to online OMIM searches, which trainees will also be expected to become familiar with)*

Emery's Elements of Medical Genetics (12<sup>th</sup> edition) Emery, Turnpenny and Ellard eds. (2005) Churchill Livingstone

**MOLECULAR GENETICS**

Human Molecular Genetics (4<sup>th</sup> edition) Strachan and Read (2010) Garland Science Publishers.

Introduction to risk calculation in genetic counselling. Young (1999) Oxford Medical Publications

PCR Technology. Current Innovations. Griffin and Griffin (2002) CRC Press

DNA Microarrays: A molecular cloning manual. Bowtell and Sambrook (2002) Cold Spring Harbour

Molecular Cloning: A Laboratory Manual, Vol. I, II and III (3rd edition), Sambrook, Fritsch and Maniatis (2001) Cold Spring Harbor Laboratory

Molecular Diagnosis of Genetic Disease (2<sup>nd</sup> edition), Elles and Mountford (2002) Humana Press

Molecular Biology of Cancer. Macdonald (2004) Taylor and Francis pubs.

PCR. McPherson, Quirke and Taylor (eds.), (1991), Oxford University Press

PCR 2. McPherson, Hames and Taylor (eds.) 1995 Oxford University Press

### **HUMAN DEVELOPMENT/PRENATAL DIAGNOSIS**

Before we are born: Essentials of embryology and birth defects. Moore and Persaud (2003), Elsevier

The Malformed Fetus and Stillbirth: A Diagnostic Approach. Winter, Knowles, Bieber and Baraitser (2000) John Wiley

Chorion Villus Sampling. Liu, Symonds and Golbus, eds. (1987) Chapman and Hall

### **CONSTITUTIONAL CYTOGENETIC INVESTIGATION/MICROSCOPY**

Hammerton. 2000. *Human Cytogenetics Vols. I & II*. Academic Press.

Human Chromosomes: Structure, Behaviour, and Effects. Therman (2001) Springer-Verlag

Human Cytogenetics: Malignancy and acquired abnormalities. Rooney and Czepulkowski, eds. (2001) Oxford University Press

Catalogue of Unbalanced Chromosome Aberrations in Man (2<sup>nd</sup> edition). Schinzel (2001) Walter de Gruyter

An International System for Human Cytogenetic Nomenclature (ISCN) 2009 Ed L G Shaffer, M L Slovak, L J Campbell

Light Microscopy in Biology: A Practical Approach. Lacey, ed. (1989) IRL Press

Human Cytogenetics vol I Constitutional Analysis (2<sup>nd</sup> edition) Rooney and Czepulkowski (1992) Oxford University Press

Chromosome Abnormalities and Genetic Counselling, Gardner and Sutherland 3rd Ed 2004

Catalogue of Unbalanced Chromosome Aberrations in Man Schinzel 2<sup>nd</sup> Ed 2001

### **OTHER SUBJECTS**

Dorland's Illustrated Medical Dictionary. Taylor, ed. (2000) Saunders

Essential Haematology (4<sup>th</sup> edition). Hoffbrand and Pettit (2001) Blackwell

Essential Immunology (10<sup>th</sup> edition). Roitt (2001) Blackwell

## **HEALTH AND SAFETY/DATA PROTECTION**

The Management of Health and safety at Work Regulations (1999), HMSO

The COSHH Regulations (2002), HMSO

Categorisation of Pathogens According to Hazard and Categories of Containment, Advisory Committee on Dangerous Pathogens (1984) HMSO

Safety in Health Service Laboratories (Hepatitis B), Health Services Advisory Committee (1985) HMSO

Safety in Health Service Laboratories: the Labelling, Transport and Reception of Specimens, Health Services Advisory Committee (1986) HMSO

The Genetics manipulation Regulations (1989) HMSO

## **GENETICS WEB LINKS:**

### **Genetics databases**

OMIM- On-line Mendelian Inheritance in Man  
<http://www.ncbi.nlm.nih.gov/omim>

GeneCards  
<http://bioinformatics.weizmann.ac.il/cards/>

GeneClinics  
<http://www.geneclinics.org/>

Human Gene Mutation Database (HGMD)  
<http://www.hgmd.cf.ac.uk/ac/index.php>

Orphanet  
<http://www.orpha.net/consor/cgi-bin/index.php>

UK Genetic Testing network  
[www.ukgtn.org](http://www.ukgtn.org)

### **Aids to learning genetics / genetics information**

DNA from the Beginning  
<http://vector.cshl.org/dnaftb/>

Human Genome project

[http://www.ornl.gov/sci/techresources/Human\\_Genome/home.shtml](http://www.ornl.gov/sci/techresources/Human_Genome/home.shtml)

National Genetics Education and Development Centre

<http://www.geneticseducation.nhs.uk/>

Nature Education – Scitable

<http://www.nature.com/scitable/topics>

### **Professional organisations**

Association for Clinical Cytogenetics

<http://www.cytogenetics.org.uk>

Clinical Molecular Genetics Society

<http://www.cmgs.org/>

British Society for Human Genetics

<http://www.bshg.org.uk>

European Society of Human Genetics

<http://www.eshg.org>

Clinical Pathology Accreditation (UK) Ltd

<http://www.cpa-uk.co.uk/>

Medicines and Healthcare Products Regulatory Agency

<http://www.mhra.gov.uk/Howweregulate/Medicines/Inspectionandstandards/GoodClinicalPractice/index.htm>

### **Ethical issues**

Nuffield Council on Bioethics

<http://www.nuffield.org/bioethics/index.html>

GeneWatch UK

<http://www.genewatch.org/>

### **OTHER USEFUL SERIES/JOURNALS**

American Journal of Human Genetics

American Journal of Medical Genetics

Clinical Genetics

Genomics

Human Genetics

Human Molecular Genetics

Human Mutation

Journal of Medical Genetics

Lancet

Nature

Nature Genetics

New England Journal of Medicine

Prenatal Diagnosis

Scientific American

Science

Trends in Genetics

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