

National Healthcare Science School of Genetics

Healthcare Scientist Training Programme Clinical Laboratory Genetics

Training Manual

Modernising Scientific Careers

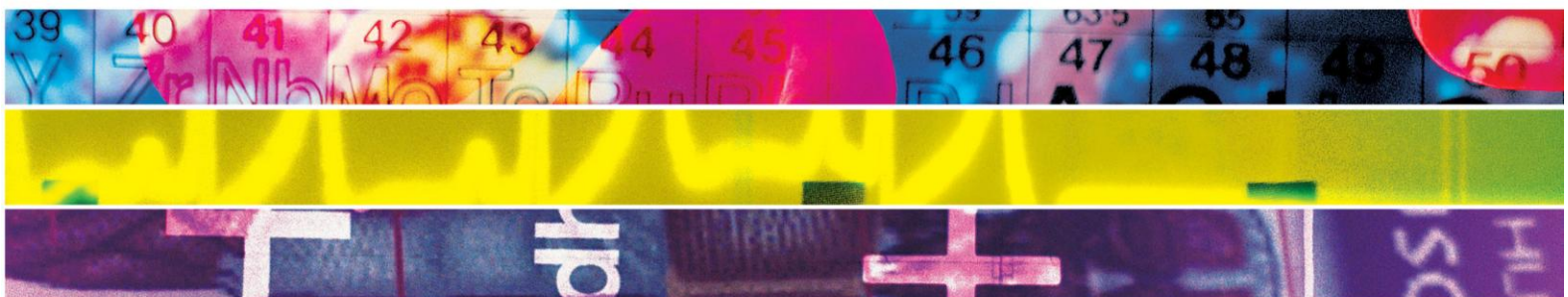


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1.0 Healthcare Scientist Training Programme (STP) in Genetics

1.1 Introduction

This manual describes the structure and the functions of the training programme for a Healthcare Scientist (HCS) in Clinical Laboratory Genetics over the next three years, the assessment strategies and your portfolio structure. This training manual must be used in conjunction with the Nottingham University MSc in Clinical Genetics course handbook, which will give details of the academic component of the training programme and the Online Assessment Manual, which will provide all the information necessary for the online assessment tool and the e-portfolio.

Any training programme must recognise the roles and functions the Healthcare Scientist is expected to successfully perform at the end the programme.

1.2 Role of a Healthcare Scientist

A Healthcare Scientist (HCS) will have clinical and specialist expertise underpinned by theoretical knowledge and experience and will;

- Undertake complex scientific and clinical roles, including those working directly with patients
- Analyse, interpret and compare investigative and clinical options
- Make judgements, including clinical judgements involving complicated facts or situations that impact on patients
- Initiate and undertake innovation, improvement and R&D, be involved in the education of trainees and other learners in the workplace

Scientific interpretation following technical analysis in a range/combination of: -

- Molecular testing – for example gene sequencing, fragment analysis
- Gene chips – for example resequencing
- Molecular cytogenetics – for example microarrays and FISH
- Chromosome analysis
- Molecular pathology
- Oncology
- Utilisation of genomic databases/bioinformatics
- Detailed data analysis
- Clinical and scientific input into specialised services
- Involved significantly in the day-to-day leadership of parts of the service to nationally accepted standards (CPA or equivalent)

More detailed and generic role descriptors can be found in **Appendix 1**.

1.3 Good Scientific Practice

Good Scientific Practice sets out the principles and values on which good practice undertaken by Healthcare Scientists is founded.

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 2 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.

1.4 Overall Aim of the Training Programme

The overall aim of this HCS training and education programme is to prepare the trainee to fulfil the function of a genetics scientist working in a clinical healthcare setting.

The programme has been designed not only to combine both the traditional disciplines of molecular and cytogenetics but also to have a strong patient and clinical focus.

1.5 Module Design

The modules have been designed around specific patient pathways which are currently pertinent to genetic testing, but with an eye to the developing technologies and their impact on future service delivery. This training programme will evolve as patient and service needs change.

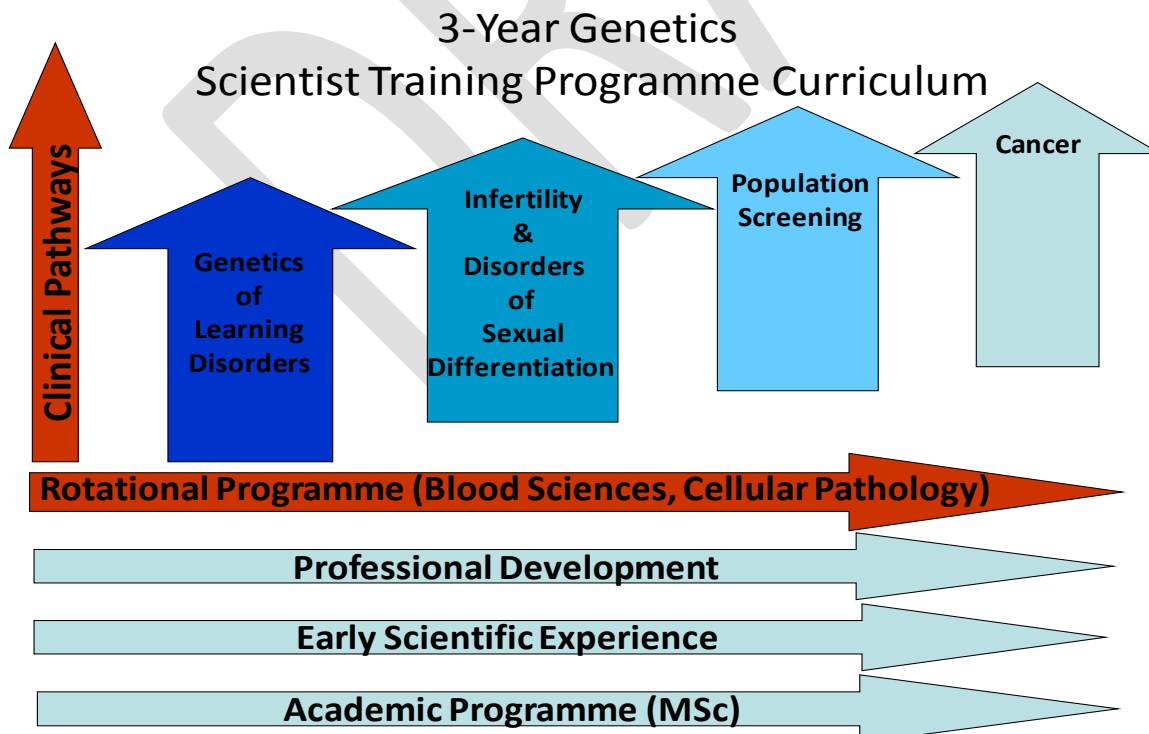
The style of the learning outcomes is such that future roles can easily be introduced into the programme.

The clinical modules are work based with continuous assessment. 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.

The modules are designed in such a way as to build up practical and analytical skills as the trainee progresses throughout the programme rather than to have separate and artificially distinct cytogenetic and molecular genetic modules. Within the current module design, the practical skills will move from chromosome to gene analysis.

The University of Nottingham is the academic partner for this programme and a significant part of the theory including genetics and healthcare and disease diagnosis, will be delivered by them in national courses. A significant component of the generic curriculum will be delivered in the formal University setting.

Figure 1 Healthcare Scientist Training Programme Curriculum



The University of Nottingham will also deliver some of the training on future technologies e.g. second-generation sequencers, but experiential learning will also be included where possible and appropriate.

Other external and additional specialist teaching will be brought in as necessary e.g. genetic risk assessment and bioinformatics.

Each clinical module will have appropriate patient interaction, which will be organised locally.

1.6 Joint meeting with Healthcare Science Practitioner Trainees (HCSP)

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:

1.6.1 Indicative Content of Course

- Introduction to MSC
 - Career development framework
 - Exciting developments across health care
- 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

2.0 The Training Programme

2.1 Pathology Rotation

The practical arrangements for the pathology rotations will need to be organised locally but it will be helpful to the trainees if they are able to follow the general outline of the syllabus as set out below.

Whatever the local arrangements, all rotations must be complete within the first two years of the programme to allow the final year for specialisation and research within genetics. The rotational programme for genetics has now been agreed for other disciplines.

2.2 Academic Course Structure

The trainees will begin in early October 2010 in their own laboratory

A national course organised by the National Healthcare Science School of Genetics will introduce the role of HCS in Genetics. This will be a one-day course held in conjunction with the Healthcare Science practitioners

2.2.1 Content of the first course for the Msc In Clinical Genetics; Introduction to Healthcare Science

Module aims:

This module aims to provide trainees with introductory knowledge and understanding of the core clinical subjects within the Modernising Scientific Careers Healthcare Scientist Clinical Genetics training programme. The module will also highlight key attitudinal and professional attributes and establish a framework for their future development, as well as provide an overview of the main teaching and learning methods adopted for the programme.

Intended learning outcomes:

Trainees will gain knowledge and a basic understanding of the following:

Introduction to Clinical Science

The cardiovascular system

- Anatomy and physiology of the heart
- Blood vessels and transport of blood
- Major vasculature

Haematology and transfusion science

- blood as a tissue
- blood cells (types and life times)
- blood cell production and blood counts
- blood groups and their genetics

Clinical Immunology in health and disease

- Innate immunity, interferon, cytokines and chemokines
- Humoral immunity
- Cell mediated immunity
- HLA types and their consequences
- Principles of autoimmune disease

The respiratory system

- Anatomy and physiology of the lung
- Ventilation
- Gaseous exchange and blood gas transport

Nerves and muscle: An introduction to excitable tissues

- Basic organisation of the central and peripheral nervous system.
- Module Objectives
- Know: To describe the basic structure and function of the peripheral and autonomic nervous system
- Intellect: to demonstrate effective use of opportunities for self-directed and independent learning.
- Transfer: To use library and other resources for self-directed learning
- Key functions of the nervous system.
- Understand the physiology and mechanism of neural transmission.
- Provide an overview of skeletal and smooth muscle function and activation

The GI tract

- Normal structure and function of the GI tract
- Digestion and absorption of food
- Liver and liver function tests

The renal and endocrine systems

- The normal structure and function of the kidney
- Electrolyte and acid-base balance
- Anatomy and function of the endocrine system
- Overview of hormonal mechanisms and control

The reproductive system

- Overview of sexual differentiation, including differentiation of the foetal testes and ovary, and the endocrinology and embryology of sexual differentiation
- The anatomy and physiology of the male reproductive tract
- The anatomy and physiology of the female reproductive tract

Introduction to cell biology and genetics

Eukaryotic cell biology

- Cell structure
- Function of the major organelles
- Nucleus, sER/rER, Golgi, endo-/exocytotic vesicles, proteasome
- Cell division, meiosis, mitosis and chromosome segregation
- Nucleic Acid structure and function, chemical structure of DNA and replication, transcription, and translation
- Gene structure and function genome variants
- Amino acid structure and function, physical attributes

Genetics in health and disease

- Chromosomes – structure and function
- Chromosomal abnormalities - mechanisms and origin of numerical and structural abnormalities, and behaviour of structural chromosome anomalies at meiosis
- Patterns of inheritance – autosomal dominant and recessive, X-linked
- Genetic basis of acquired disease / Cancer
- Genetics
- Genomics
- Pharmacogenetics
- Bioinformatics
- Clinical Genetics
- HGVS and ISCN nomenclature

Healthcare Science

Being a Scientist (V)

- Scientific skills
- Legal Framework for practice
- Good Laboratory Practice
- Good Scientific Practice
- Communication with colleagues and cooperation
- Time management and organisation

- Leadership and Management (MLCF)
 - Demonstrating Personal Qualities
 - Working with Others
 - Managing Services
 - Improving Services
 - Setting Direction
- Informatics
- Continuing Professional Development including Reflective Practice

Introducing Quality Assurance (V)

- Quality control, assurance, Laboratory and clinical accreditation
- Quality improvement, metrics and benchmarking
- Clinical governance

Developing Clinical/Diagnostic/Investigative Skills (V)

- History taking
- Differential diagnosis and role of assessment and investigation
- Appropriateness and predictive value of diagnostics
- Presenting symptoms and signs
- Record keeping and documentation
- Handling uncertainty
- Consultation skills including:
 - Key features of effective patient interviews and information giving
 - Illness behaviour, scientist and patient roles and relevant cultural beliefs
 - Awareness of the key features of effective patient interviews and information giving
 - Communication skills with groups of the population who have particular communication needs such as children, those with learning disabilities
 - Management of emotional responses within the scientist-patient interaction
- Risk probability
- Care models and patient pathways of care
- Informatics
- Clinical coding/terminology
- Clinical Information systems and applications
- Data protection/Caldicott

(V) Indicates vertical learning themes that will be delivered throughout the MSc programme, particularly during problem based learning sessions.

Assessment methods:

Achievement of the intended learning outcomes will be assessed via a 1.5 hour MCQ/short essay paper and a 1,000 word assignment.

National MSc in Clinical Genetics– weeks and location to be confirmed by The University of Nottingham **for details and dates refer to the course hand book**

2.3 Generic Learning

There are a number of generic competencies which arise out of Good Scientific Practice and which focus on attitudes, behaviours, knowledge and skills. These map to the professional standards needed for registration with the appropriate regulatory bodies. These will be delivered throughout the programme and include areas such as communication and Health and Safety which will complement and test application of the academic learning. These will be assessed throughout the work based assessment programme and will be demonstrated using the assessment tools DOPs and CBDs (see section 5). There are competences for the leadership and management part of this in the competencies log book and these again should be filled in when appropriate throughout the training. The CBDs and DOPs will both identify areas of competence in leadership and management.

2.3.1 Associated Personal Qualities/Behaviours/Professionalism

The scientist clearly has to operate to high standards of professionalism and demonstrate essential behaviours and personal qualities. These are developed and assessed in accordance with the following learning outcomes.

Learning Outcomes

1. Present complex ideas in simple terms in both oral and written formats
2. Consistently operate within sphere of personal competence and level of authority
3. Manage personal workload and objectives to achieve quality of care
4. Actively seek accurate and validated information from all available sources
5. Select and apply appropriate analysis or assessment techniques and tools
6. Evaluate a wide range of data to assist with judgements and decision making
7. Conduct a suitable range of diagnostic, investigative or monitoring procedures with due care for the safety of self and others
8. Report problems and may take part in restorative action within quality control/assurance requirements to address threats of performance deterioration
9. Work in partnership with colleagues, other professionals, patients and their carers to maximise patient care
10. Understand the legal framework within which healthcare is provided in the UK and/or devolved administrations in order to ensure that personal clinical practice is always provided in line with this legal framework.
11. Understand and demonstrate the use of a range of IT platforms/software appropriate to the role.
12. Recognise the causes of error and how to learn from them, realising the importance of honesty and effective apology.
13. Undertake research within current ethical and governance guidelines, disseminates the research output and uses own and scientific literature to continually review and develop good scientific practice.
14. Performs audit of scientific practice and apply the findings appropriately and complete the audit cycle within current ethical and governance regulations.

2.3.2 Management and leadership

Aim

To prepare the trainees for effective leadership and managerial responsibilities in their role as HCS.

1. Recognises and accepts the responsibilities and role of the scientist in relation to other healthcare professionals.
2. Communicate succinctly and effectively with other professionals as appropriate offering both professional and clinical advice.
3. Demonstrates increasing ability to prioritise and organise duties in order to optimise the work of the department. Demonstrates improving ability to make appropriate decisions in order to optimise the effectiveness of the scientific team resource.
4. Demonstrates the skills and experience to undertake effective, appropriate leadership and management responsibilities in a genetics laboratory.
5. Teaches a range of health professionals and other audiences in a variety of different ways and assess the quality of the teaching of self and others.
6. Understands the structure of the NHS and the management of local healthcare systems in order to be able to contribute appropriately in managing genetics services.

Indicative content

- Leadership and management theories
- Theory of change management
- Clinical governance
- Audit
- Team building and working theories
- Attending senior laboratory management meetings

3.0 Clinical Laboratory Genetics Training

3.1 MODULE 1 Genetics of Learning Disorders

3.1.1 Aim

To understand the role and application of genetics testing in the diagnosis and management of patients with learning difficulties and the implications for other family members.

This module is work based but is underpinned by a national course. The indicative content of the national course is set out in the **Nottingham Course Handbook**.

The practical skills for the module are indicated below with the detail provided in the **Competencies in section 5**.

During this module it is intended that the trainee will have considerable practical experience and competence in carrying out chromosome analysis and array analysis and they will experience and understand the molecular technology.

3.1.2 MSc in Clinical Genetics to be delivered by the University of Nottingham

Course Content

- Clinical presentation, natural history and modes of inheritance of syndromes and disorders associated with learning difficulties using chromosome abnormalities, fragile X syndrome, Prader Willi and Angelman (PWS/AS) syndromes, and Rett syndrome as examples
- Clinical and scientific aspects of chromosome disorders
- Telomeric deletions – telomeric structure
- Microdeletion syndromes and chromosome architecture: genomic disorders
- Dysmorphology and the underlying genetics
- The clinical scientific and molecular basis of imprinting disorders with particular reference to PWS/AS syndromes
- X-linked mental retardation: the clinical scientific and molecular basis of fragile X syndrome and Rett syndrome
- The clinical utility of genetic testing in the above conditions for a range of common clinical scenarios (for example diagnostic testing, carrier testing, identification of normal transmitting males in fragile X, prenatal diagnosis)
- Use of pedigree analysis to determine the most appropriate family member for testing for any given referral reason for chromosomal disorder, fragile X, PWS/AS and Rett syndrome
- Social benefits of a positive diagnosis in learning disability (educational statements of need, adoption)
- Databases and patient support: DECIPHER and Unique
- The patient and family perspective of securing a diagnosis, understanding future prognosis and risk
- Clinical care pathways associated with genetic testing for learning disorders
- Array technology and analysis
- Theory behind basic techniques used in this module – chromosome analysis, arrays, sequencing Ethical issues associated with consent and DNA testing

3.1.3 Work Based Learning Outcomes

Learning outcome 1

Demonstrate an understanding of a range of genetics testing relevant to learning difficulties

using, chromosome analysis, micro-array analysis, fragile X syndrome, Rett syndrome, PWS/AS as examples.

Learning outcome 2

To demonstrate the ability to develop an appropriate testing strategy for learning difficulties.

Learning outcome 3

Demonstrate the ability to perform chromosome analysis.

Learning outcome 4

Demonstrate the ability to perform microarray analysis.

Learning outcome 5

Demonstrate an ability to perform a basic analysis and interpret fragile X syndrome, Rett syndrome, PWS/AS.

Learning outcome 6

Demonstrate an understanding of the implications of the genetics tests including ethical, legal and social implications for the effective management of the patients.

3.1.4 Indicative content and practical skills to be delivered in the work place

- The principal referral reasons which would indicate testing for each of the conditions under investigation.
- Blood sample processing to obtain chromosome preparations suitable for analysis and appropriate staining and banding techniques relevant to this referral group.
- Analysis of chromosomes from patients with learning disorders – use of ISCN.
- Interpretation of results from chromosome analysis using relevant on- line databases and search engines.
- Application of FISH for targeted micro deletion/duplication syndromes: technical processing and fluorescent microscopy.
- Assessment of suitability of DNA for microarray analysis.
- Microarray technical processing: labelling, hybridisation, scanning.
- Microarray data-analysis and using array-CGH software.
- Interpretation of the significance of Copy Number Variations (CNVs) using genome browsers and CNV databases and reporting using ISCN.
- Design of strategies for extended microarray follow-up using additional techniques.
- The clinical, scientific and molecular basis for the repertoire of genetic testing available to investigate the common range of clinical referrals for fragile X syndrome, syndrome and PWS/AS syndrome.
- Interpretation of results from fluorescent PCR across the FMR1 gene in order to detect results within normal, intermediate and expanded ranges, and results, which are to be designated as failed analyses. Identification of samples which require repeating and/or which require Southern blotting.
- Interpretation of results from Southern blotting for the detection of large expansions and

methylation status of alleles in Fragile X.

- Interpretation of results from MLPA or bisulphite analysis for PWS/AS syndrome.
- Use of Southern blotting to detect methylation differences in PWS/AS syndromes.
- Applicability of non-routine investigations available to elucidate unusual results further (for example sequencing of FMR1 gene).
- Understanding of LIMS to access correct reporting templates.
- Significance of test results and interpretation in order to inform further testing to answer clinical questions.
- Sources of further information for extended testing in learning disability e.g. use of UK Genetics Testing Network (UKGTN) and DECIPHER databases to identify commissioned tests and where they could be undertaken.

3.1.5 Suggested Clinical Experience – these are suggestions but may vary according to local implementation

- Attend genetics dysmorphology counselling clinics
- Attend school for children with learning disabilities
- Ward rounds with consultant geneticist at a local children's hospital
- Discuss nutritional needs of PWS etc with dietician

3.2 MODULE 2 Infertility and Disorders of Sexual Differentiation

3.2.1 Aim

To understand the role and application of genetics testing in the diagnosis and management of patients with infertility and disorders of sexual differentiation and the implications to other family members.

This is the second work based module and is designed to look at the disorders of sexual development and infertility.

The main practical element of this module will be to become familiar and competent with molecular testing for Cystic Fibrosis (CF) and use this as an example for testing for other single genes disorders and looking at the implications of other CF mutations and different patterns of referral for CF testing.

However the trainee will also maintain their competence in chromosome analysis by examining samples from patients with infertility and sexual disorders.

3.2.2 MSc in Clinical Genetics to be delivered by The University of Nottingham

This module will also be underpinned by a national course, which will either take place alongside this module or in combination with another national course

Course Content

- Basic embryology and normal sexual differentiation.
- Range of phenotypes and genotypes associated with disorders of sexual development.
- Patterns and causes of abnormal sexual differentiation, both autosomal and sex chromosome related.
- The major genes associated with normal sexual differentiation.
- The genetic basis of infertility and options for management.
- Ethical, legal and social issues of individuals and families affected by disorders of sexual differentiation.

3.2.3 Work Based Learning Outcomes

Learning outcome 1

Demonstrate an understanding of a range of genetic testing including chromosome analysis, CF testing, Y deletion screening, androgen receptor mutation analysis, FRAXA testing in premature ovarian failure (POF), relevant to infertility and disorders of sexual differentiation.

Learning outcome 2

To demonstrate the ability to develop an appropriate testing strategy relevant to infertility and disorders of sexual differentiation.

Learning outcome 3

Demonstrate the ability to perform CF testing, FRAXA and chromosome analysis relevant to sexual differentiation and other disorders.

Learning outcome 4

Demonstrate the ability to interpret and report a range of genetic testing relevant to infertility and disorders of sexual differentiation including abnormal results from chromosome constitution, CF, Y deletion, Androgen receptor, POF, sex reversal.

Learning outcome 5

Demonstrate an understanding of the implications of the genetic tests including ethical, legal and social implications for the effective management of the patients.

Learning outcome 6

Understands the necessity of obtaining valid consent from the patient, knowing how to obtain it when it is indicated.

Learning outcome 7

Accurately record a family history and synthesise this with relevant clinical information including differential diagnosis(es) to formulate a management plan that takes account of likely clinical evolution.

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

- Review of testing strategies appropriate to this group of referrals and relevant patient pathways
- Recognise problems associated with genetic mosaicism in testing this group of patients.
- Relevant laboratory techniques used to identify genomic abnormalities in this group of patients
- The technical basis of the major genetic tests (e.g. ARMS and OLA) to detect CFTR mutations including intron 8 polyT tract variants.
- The technical basis of detecting FRAXA premutation alleles in POF referrals by PCR and Southern blot analysis.
- Comparative advantages and disadvantages of each method for detecting FRAXA premutations.
- Relationships between particular genetic abnormalities and their influence on phenotype.
- Diagnostic and prognostic significance of genetic abnormalities found in this group of patients
- Recognise and understand this type of genetic testing in relation to other clinical referrals and laboratory investigations.

3.2.5 Suggested Clinical Experience - these are suggestions but may vary according to local implementation

- Attendance at infertility clinics
- CF clinics
- Assisted conception unit visit
- Attendance at paediatric endocrine clinic/ward round

3.3 MODULE 3 Population Screening

3.3.1 Aim

To understand the role and application of genetic testing in population screening and the associated diagnosis and management of patients.

This module has been designed to give a wider context of genetic testing and its utility following from specific screening programmes; the role of new screening programmes e.g. familial hypercholesterolemia (FH), fDNA and RNA and their likely impact on genetics departments in the future.

The main practical skills will focus on QF-PCR and FISH testing for aneuploidy detection and chromosome and array analysis following an abnormal Ultrasound Scan (USS), and targeted mutation screening. Where possible there should also be experiential learning of fDNA/RNA technologies.

3.3.2 MSc in Clinical Genetics to be delivered by The University of Nottingham

Course Content

- The role of screening in the delivery of the Public Health agenda.
- Principles of screening and the different approaches including the relationship between the genetic testing and other screening modalities such as Ultrasound Screening (USS) and Immunoreactive Trypsin (IRT) across a range of disorders such as MCAD, CF, and PKU.
- Review of relevant issues such as compliance and choice, cost effectiveness and clinical pathways.
- Principles of population screening including concepts of sensitivity, specificity and positive predictive value.
- Origin, prevalence and clinical significance of the most commonly encountered chromosome abnormalities and genetic disease encountered in prenatal diagnosis following screening programmes.
- Associations between various abnormal ultrasound scan signs and chromosome abnormalities.
- Conditions screened in the national newborn screening programmes and the role of genetic testing in screening for these conditions for example Haemoglobinopathies.
- Future screening programmes – for example cardiac screening.
- Principles and technologies underpinning non-invasive prenatal diagnosis and its role in future screening programmes.
- Ethical, legal and social implications for population screening.
- Guidelines/models of best practice for screening (such as WHO guidelines) and the role of the National Screening Committee.

3.3.3 Work Based Learning Outcomes

Learning outcome 1

Demonstrate an understanding of a range of genetic testing relevant to population screening including prenatal screening and following diagnostic testing using as examples Sickle cell/Thalassaemia, CF, PKU, MCAD, Down syndrome screening, FH, USS and an understanding of non-invasive prenatal diagnosis.

Learning outcome 2

To demonstrate the ability to develop an appropriate genetic testing strategy relevant to diagnosis following population screening.

Learning outcome 3

Demonstrate the ability to perform, QF-PCR and interphase FISH for trisomy detection, high resolution analysis of chromosomes and targeted mutation analysis to nationally agreed standards.

Learning outcome 4

Demonstrate the ability to interpret and report a range of genetics testing relevant to the

effective management of the patient, in relation to the national screening programmes and public health.

Learning outcome 5

Demonstrate an understanding of the implications of the genetic testing including ethical legal and social implications for the effective management of the patients.

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

- Targeted mutation testing and aneuploidy testing without generating unwanted information.
- Best practice guidelines when involved in a complex and/or tightly defined care pathway.
- Principles and practice of QF-PCR used for aneuploidy detection and in relation to other uses of this technology.
- Nature and effect of structural chromosome rearrangements in relation to QF-PCR testing.
- Nature and effect of possible artefacts related to QF-PCR.
- The nature and effect of mosaicism, maternal cell contamination, twin pregnancies in relation to QF-PCR testing.
- Procedures for confirmation of abnormal QF-PCR results.
- Principles and practise of interphase FISH and the utility of FISH vs QF-PCR.
- ACC/CMGS Best Practice Guidelines relevant to analysis of QF-PCR for diagnosis of aneuploidy.
- Utility of high resolution chromosome analysis / array analysis following abnormal USS.
- Methods of targeted mutation analysis and their use in screening protocols such as ARMs. OLA, Pyrosequencing.
- The integration of targeted mutation analysis associated with screening protocols with genetic testing for the same disease in other care pathways.
- The role of genetic testing in the screening programmes, the information that is required to deliver the screening outcomes and therefore the content of the report.
- The clinical implications of any of the results from the range of genetic tests.

3.3.5 Suggested Clinical Experience - these are suggestions but may vary according to local implementation

- Observation of abnormal ultrasound scans
- Observation of invasive prenatal procedure
- Attendance at special fetal medicine genetics clinic
- Haemoglobinopathies clinic

3.4 MODULE 4 Cancer

3.4.1 Aim

To understand the role and application of genetic testing in the diagnosis and management of patients with acquired and inherited cancer and the implications to other family members.

This module is designed to give a wide understanding of selected acquired and inherited cancers and to develop an understanding of the role of genes involved and how they can be analysed. Various cancers will be used as specific examples to demonstrate the underlying principles of oncogenesis and the practice of genetic testing in diagnosis, prognosis and management.

3.4.2 MSc in Clinical Genetics to be delivered by the University of Nottingham

Course content

- Cell cycle regulation.
- Gene pathways associated with cancer.
- Principles of carcinogenesis, oncogenesis and malignant transformation.
- Proto-oncogenes, Oncogenes and tumour suppressor genes.
- Common translocations and rearrangements associated with cancer initiation and progression.
- Common genetic mutations associated with cancer initiation and progression.
- Knudson two hit hypothesis.
- Patterns of inheritance associated with predisposition to cancer including the issue of genetic heterogeneity, environmental factors and multifactorial inheritance (including awareness of chromosome instability syndromes).
- Principles of high throughput sequencing and its application to screening large genes
- Haematopoiesis with particular reference to stem cell theory and classification.
- Selection and analysis in mixed cell populations.
- The concept of Minimal Residual Disease (MRD) and its utility in disease treatment and monitoring.
- Transplantation and chimerism monitoring.
- Pharmacogenetics and personalised treatment.
- Ethical, legal and social implications of genetic testing in cancer.

3.4.3 Work Based Learning Outcomes

Learning outcome 1

To demonstrate the ability to develop an appropriate testing strategy for acquired and inherited cancers, such as BRCA, HNPCC, CML, ALL including molecular pathology, emerging bio-markers and methylation including risk assessment.

Learning outcome 2

Demonstrate the ability to perform mutation scanning and sequencing of large genes such as the inherited cancer genes, FISH for the detection of cancer fusion genes, quantitation, and chromosome analysis of leukaemia.

Learning outcome 3

Demonstrate the ability to interpret and report a range of genetic testing relevant to cancer including acquired and inherited, predictive testing, pedigree analysis, BRCA, HNPCC, molecular pathology and bio-markers.

Learning outcome 4

Demonstrate an understanding of the implications of the genetics tests including ethical, legal and social implications for the effective diagnosis and prognostic management of the patients.

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

- Clinical care pathways associated with the samples tested in cancer and the principles of cost effectiveness.
- Ethical issues associated with consent including predictive testing.
- Basis of inherited predispositions to cancer using examples of two diseases such as BRCA, FAP or HNPCC.
- Principles of pedigree analysis and the calculation of risk.
- Principles and practice associated with predictive testing in inherited cancer syndromes.
- Principles underpinning at least two methods of mutation scanning including sequencing.
- Genetic causes of sporadic cancer such as sporadic bowel cancer, the gene pathways involved and their relation to inherited disease.
- Gene structure and the range of mutations seen in an exemplar large gene.
- Different methodologies that can be used to identify different classes of mutation.
- The scientific and mathematical basis of quantitation using standard curves, regression analysis and relevant software.
- Use of biomarkers such as somatic mutations such as those found in KRAS in the management and treatment of cancer.
- Basis upon which variants identified in the germline are classified according to their pathology.
- Utility of genetic testing in generating prognostic information in the management of cancer e.g. common chromosomal rearrangements.
- Utility of genetic testing in monitoring the efficacy of treatment in cancer e.g. CML, Breast cancer.
- Role of managerial processes such as Multidisciplinary Team (MDT) meetings and guidelines such as Improving Outcomes Guidance and NICE Guidelines.
- Relationship between chromosome abnormalities and oncogenes and an understanding of the relationships between chromosome abnormality and the molecular biology of cancer.
- Role of cytogenetics in bone marrow transplantations.

- Role of molecular analysis in the diagnosis and monitoring of leukaemias.
- Mixed cell populations seen in cancer and how the testing strategy has to be developed and refined if required to enhance the population of abnormal cells.
- Principles of FISH in identification of rearrangements associated with cancer.
- Principles underpinning at least one method in quantitation of residual disease e.g. CML, ALL.
- Rearrangements and translocations commonly associated with cancer, their clinical significance and the methods used to detect them.
- Other methods used in the diagnosis of leukaemias (i.e. haematology, morphology, immunology).

3.4.5 Suggested Clinical Experience - these are suggestions but may vary according to local implementation

- MDT meetings
- Observing bone marrow aspiration
- Attending cancer genetics counselling clinic
- Attending haematology clinic

3.5 MODULE 5 Research Project carried out in conjunction with The University of Nottingham

3.5.1 Aim

To enable the student to gain the skills and knowledge in research methods and apply these skills by identifying an appropriate research question, testing a hypothesis, presenting the results and understanding the limitations and applicability of their research to the NHS.

3.5.2 Work Based Learning Outcomes

Learning outcome 1

Demonstrate an ability to identify an appropriate area for development and its application to clinical practice.

Learning outcome 2

Demonstrate knowledge of scientific experimental design and planning of a robust scientific experiment, including awareness of health and safety, cost implications and ethical issues.

Learning outcome 3

Demonstrate the ability to conduct a scientific experiment, where there may be more than one variable or where there are two components to the experiment, the second of which is dependent on the results of the first, including problem solving, relevant data collection, analysis methods and statistical evaluation.

Learning outcome 4

Demonstrate an ability to critically evaluate the process and findings of the research and disseminate via a written report.

Learning outcome 5

Demonstrate effective time management skills and workload prioritisation in the context of scientific research and writing.

Learning outcome 6

Undertake research within current ethical and governance guidelines, disseminates the research output and uses own and scientific literature to continually review and develop good scientific practice.

Learning outcome 7

Makes optimal use of current best evidence in making decisions and develops the ability to construct evidence based guidelines and protocols in relation to scientific practise.

Learning outcome 8

Performs audits of scientific practice and apply the findings appropriately and complete the audit cycle within current ethical and governance regulations.

3.5.3 Indicative content to be delivered in the workplace in conjunction with The University of Nottingham

- Searching and reviewing literature
- Experimental design
- Research methodologies
- Data analysis tools
- Statistics
- Health and Safety
- Ethics

3.6 Rotational Element

The rotational element of the genetics pilot will comprise of three, 12 week placements within other life science disciplines, interspersed over the first two years.

The teaching and learning strategy for these rotational programmes will be based upon case study and approach to learning. The assessment will follow the MSC assessment strategy format described above for genetics training.

**3.6.1 Clinical Biochemistry Rotation
Investigation of Major Organ Function**

This module will provide trainees with the knowledge and understanding of the normal physiology of the major organs and the biochemical parameters in common use for the

investigation and management of major organ dysfunction. They will perform common methods used in the investigation of major organ function and gain experience of the interpretation of patient results in a variety of clinical settings.

Academic Knowledge and Understanding

Learning Outcomes

On successful completion of this module the student will be able to:

1. Understand normal physiological homeostasis of the major organs
2. Understand the of pathophysiology and cause of common disorders of the major organs
3. Explain the presentation, diagnosis and management of common biochemical disorders of major organ function
4. Understand the principles of common biochemical measurement techniques used to investigate major organ function
5. Describe the design, operation and performance of automated analytical platforms used to investigate major organ function
6. Describe the design, operation and performance of point of care testing devices supported by the clinical biochemistry laboratory
7. Understand the biochemical investigation of major organ disease in the patient pathway, the correct sampling technique and the use and validity of reference ranges
8. Understand the principles of internal quality control and external quality assessment
9. Understand the use of laboratory IT systems for handling, processing and storage of patient data
10. Describe the linkages of clinical biochemistry to other clinical specialisms in the investigation of disorders of major organs

Associated Work Place Curriculum

High level description of the work place based learning that accompanies this academic module. Further details are contained within the work place training manual

Learning Outcomes

On successful completion of this module the student will be able to:

1. Gain experience of the biochemical investigation of major organ disease in the patient pathway, the correct sampling technique and the use and validity of reference ranges
2. Understand and demonstrate the ability to perform the range of laboratory and point of care techniques used in the work place to investigate major organ function
3. Demonstrate the ability to apply the principles of internal quality control and external quality assessment and draw conclusions about assay performance
4. Demonstrate the ability to use laboratory IT systems for handling, processing and storage of patient data
5. Gain experience of the interpretation and reporting of laboratory results in the context of common clinical disorders
6. Gain experience of the linkages of clinical biochemistry to other clinical specialisms in the

investigation of disorders of major organs

Indicative Content:

- The normal physiology and function of the following major organs: kidney, liver, heart, lungs, bone and pancreas. To include water homeostasis and acid base balance.
- The clinical and scientific basis of common biochemical markers of function of the kidney, liver, heart, lungs, bone and pancreas
- The application of common biochemical markers of major organ function to a range of frequently encountered clinical disorders
- Presentation, diagnosis and management of common clinical biochemical disorders of major organ function
- The biological and statistical basis of biological variation, reference values and action limits
- Principles and practice of internal quality control and external quality assessment
- Scientific basis of the following techniques: spectrophotometry, osmometry, ion selective electrodes, enzymology, immunochemical techniques, electrophoresis, chromatography, and solid phase chemistry
- Design, operation and performance of automated analytical platforms, including random access, modular, robotics etc
- Design, operation and performance of point of care testing devices supported by the clinical biochemistry laboratory

3.6.2 Haematology and Transfusion Science Rotation
Introduction to Haematology and Transfusion Science

This module will provide trainees with the knowledge and understanding of the formation of red blood cells, the mechanism of haemostasis and the relevance of blood group antigens and antibodies. They will understand the principles and practice of common methods used in haematology, haemostasis and blood transfusion and perform some of them in the laboratory. They will understand common clinical disorders associated with abnormal haematology and haemostasis and gain experience of the interpretation of patient results in a variety of clinical settings. They will gain knowledge of blood transfusion in a variety of settings, and understand how to provide patients with safe and effective transfusion support

Knowledge and Understanding

Learning Outcomes

On successful completion of this module the student will be able to:

1. Understand the haemopoietic pathways
2. Describe the design, operation and performance of the routine tests used in screening and investigating haematological disorders and their normal limits
3. Understand normal haemostatic mechanisms, and disorders causing bleeding or thrombosis
4. Describe the design, operation and performance of the tests used to investigate

disorders of haemostasis

5. Understand the concept of blood groups and the application of blood group serology in establishing compatibility between patient and donor.
6. Describe the design, operation and performance of the tests and procedures required to enable selection of safe and appropriate blood and blood components for patients.
7. Know the range of blood components and products in common use and understand the importance of correct storage
8. Describe the linkages of haematology and transfusion science to other clinical specialisms in the investigation and management of common disorders

Associated Work Place Curriculum

High level description of the work place based learning that accompanies this academic module. Further details are contained within the work place training manual

Learning Outcomes

On successful completion of this module the student will be able to:

1. Gain experience of the investigation of basic haematological disorders, the correct sampling technique and the use and validity of reference ranges
2. Understand and demonstrate the ability to perform the range of laboratory techniques used in screening and investigating haematological disorders
3. Gain experience of the investigation of basic disorders of haemostasis
4. Understand and demonstrate the ability to perform the range of laboratory and point of care techniques used in the investigation of disorders of haemostasis
5. Demonstrate the ability to perform blood group serology
6. Understand and demonstrate the ability to perform the tests and procedures required to enable selection of safe and appropriate blood and blood components for patients
7. Gain experience of the range of blood components and products in common use and their correct storage
8. Demonstrate the ability to apply the principles of internal quality control and external quality assessment and draw conclusions about assay performance
9. Demonstrate the ability to use laboratory IT systems for handling, processing and storage of patient data
10. Gain experience of the interpretation and reporting of laboratory results in the context of common clinical disorders
11. Gain experience of the linkages of haematology and transfusion science to other clinical specialisms in the investigation and management of common disorders

Indicative Content:

- Normal haemopoiesis and bone marrow function in the development and differentiation of blood cells
- Normal haemostasis and its components. Role of the liver in the production of coagulation factors
- Principles, scientific basis and clinical application of commonly performed analytical procedures in haematology

- Principles and scientific basis of automated coagulation analysers and point of care instruments in the assessment of coagulation function
- Principles and scientific basis of automated cell counters and point of care instruments for numeration and identification of cellular blood components
- Point of care testing in haematology
- Presentation, diagnosis and management of common haematological disorders
- The establishment, application and limitations of biological normal reference ranges including age, ethnic and sex related reference ranges
- Bone marrow aspiration, trephine biopsy, preparation and staining techniques for the morphological identification of cells in bone marrow in normal and pathological conditions.
- Blood film preparation, staining and interpretation in normal and pathological conditions including parasites
- Principles and application of internal quality control and external quality assurance programmes
- Basic blood group systems - genes, antigens and antibodies
- Manual and automated techniques for ABO/D typing, serological cross matching, red cell phenotyping, antibody screening and identification
- Overview of blood transfusion services and range of blood components / products manufactured and their applications
- Principles of pre-transfusion testing
- Normal ranges and predictive values for pathology tests used to inform transfusion support
- Aetiology and clinical features of conditions requiring transfusion support
- Overview of legislation / guidelines relevant to blood transfusion practice

3.6.3 Clinical Immunology Rotation

Immunity and the Principles and Practise of Clinical Immunology

This module will provide trainees with an introduction to the immune system and immune responses. They will understand the organisation and delivery of a clinical immunology laboratory service. They will perform some common methods used in clinical immunology and gain an understanding of the interpretation of patient results in a variety of clinical settings.

Knowledge and Understanding

Learning Outcomes

On successful completion of this module the student will:

1. Understand the function of the immune system in health.
1. Know and understand the function of the major cells of the immune system
2. Know and understand the function of the major humoral components of the immune response
3. Understand the innate immune system
4. Understand the adaptive immune response
5. Understand the co dependence of the innate and adaptive immune systems

6. Describe the design, operation and performance of the tests and assays used within clinical immunology.
7. Describe the linkages of clinical immunology to other clinical specialisms in the investigation and management of disorders of the immune system

Associated Work Place Curriculum

High level description of the work place based learning that accompanies this academic module. Further details are contained within the work place training manual

Learning Outcomes

On successful completion of this module the student will be able to:

1. Gain experience of the investigation of the immune response, correct sampling technique and the use and validity of reference ranges
2. Gain experience of the role of the immune response in common clinical disorders
3. Understand and demonstrate the ability to perform the range of laboratory techniques used in the work place in clinical immunology
4. Demonstrate the ability to apply the principles of internal quality control and external quality assessment and draw conclusions about assay performance
5. Demonstrate the ability to use laboratory IT systems for handling, processing and storage of patient data
6. Gain experience of the interpretation and reporting of laboratory results in the context of common clinical disorders
7. Gain experience of the linkages of clinical immunology to other clinical specialisms in the investigation and management of disorders of the immune system

Indicative Content:

- Organisation and components of the immune system
 - Cellular components (lymphocytes; granulocytes; monocytes/macrophages)
 - Humoral components (antibodies/immunoglobulins; complement; cytokines)
 - Molecules of the immune system (major histocompatibility molecules class I & II; CD molecules/cell surface markers; receptor molecules; recognition molecules; adhesion molecules; effector molecules)
 - Antigen presentation
- Innate immune response (endothelial cells; neutrophils; macrophages; natural killer cells; complement)
- Adaptive immune response (antigen processing; dendritic cells; T cell responses; B cell responses; primary and secondary responses; vaccination/immunisation)
- Outcome of immune responses (immunity/immunological memory; inflammation; direct & indirect functions of antibodies; incidental tissue damage; hypersensitivity and allergy)

3.6.4 Histopathology Rotation

Introduction to Principles and Practice of Histology

This module will provide trainees with the knowledge and understanding of the principles and practice of histology as applied to clinical medicine. They will use a range of histological techniques and gain experience of interpreting results from patient investigations.

Knowledge and Understanding

Learning Outcomes

On successful completion of this module the student will be able to:

1. Understand and recognise normal the cellular morphology of specified tissues and organs and the pathobiological processes associated with them.
2. Understand and gain experience of the receipt, preparation and processing of specimens for histopathological diagnosis
3. Understand and select the appropriate demonstration technique as part of the diagnostic process.
4. Understand and gain experience of microscopical examination techniques.
5. Understand and demonstrate the application of quality assurance methodologies.
6. Gain experience of the preparation and interpretation of clinical diagnostic reports.

Indicative Content:

- Normal cellular morphology and ultrastructure of specified tissues and organ systems, including skin, building on basic anatomy and physiology
- Introduction to tissue preparation techniques
- Specimen acquisition, viability, collection and delivery
- Principles and practice of fixation
 - Principles of Specimen Dissection and block selection
 - Tissue processing and embedding techniques
 - Decalcification
 - Microtomy, cryotomy ultramicrotomy
 - Macrophotography
- Introduction to demonstration techniques and their rationale and hazards
 - Haematoxylin and eosin
 - Special stains to identify individual tissue/cellular components, e.g. connective tissues, nucleic acids, mucins, lipids, pigments
 - Histochemical techniques
 - Immunocytochemistry
 - Molecular diagnostics
 - Electron microscopy
- Microscopy principles and practice
- Quality assurance
 - Artefacts

- Basic principles of pathobiology to include inflammation, fibrosis, necrosis, hypertrophy, hyperplasia, atrophy, metaplasia and apoptosis.

3.6.5 Cytopathology Rotation

Principles and Practice of Cervical Cytology and Diagnostic Cytopathology

This module will provide trainees with the knowledge and understanding of cervical cytology and an overview of the role and limitations of diagnostic cytopathology. They will be able to recognise normal cells in cervical cytology preparations. They will gain knowledge of the cervical screening programme, the role of fine needle aspiration cytology and non-gynaecology cytology preparation techniques

Knowledge and Understanding

Learning Outcomes

On successful completion of this module the student will be able to:

1. Understand the physiology and pathophysiology of the female reproductive tract
2. Demonstrate knowledge of the appearance of normal cellular patterns in cervical cytology.
3. Demonstrate an understanding of current cervical screening programmes.
4. Understand and apply relevant techniques for non-gynaecological cytology samples

Indicative Content:

- Overview of the cervical screening programme including, aetiology, principles of screening, coverage and call and recall and failsafe
- Understanding of the role and impact of HPV vaccination and testing on the cervical screening programme
- Principles of quality assurance including IQC, EQA and audit
- The anatomy and physiology of the female reproductive tract
- Cell patterns of normal and abnormal cervical cytology samples
- Basic understanding of the use of IT systems in cytology laboratories and the interface with laboratory computer systems.
- Treatment options for CIN and cervical cancer
- Principles of liquid based cytology and imaging technologies
- The roles of staff in a cytology department: Pathologists, Biomedical Scientists, Consultant Biomedical Scientists (Advanced Practitioners), “checkers”, Medical Laboratory Assistants and Cytology Screeners
- Principles of non-gynaecological cytology preparation techniques
- The advantages and limitations of FNA cytology in the diagnosis of benign conditions and malignant disease
- The role of immunocytochemistry and molecular techniques in the non-gynaecological cytology

3.6.6 Reproductive Science Rotation

Principles and Practice of Reproductive Science and Diagnostic Semen Analysis

This module will provide trainees with the knowledge and understanding of the normal physiology of the male and female reproductive tracts. They will be able to perform and interpret results from diagnostic semen analysis. They will gain knowledge of current legislation and regulations.

Knowledge and Understanding

Learning Outcomes

On successful completion of this module the student will be able to:

1. Demonstrate understanding of male and female reproductive anatomy.
2. Demonstrate an understanding of male and female reproductive physiology.
3. Demonstrate an understanding of current legislation and regulation as it relates to reproductive science
4. Understand and apply relevant techniques for semen analysis and preparation

Indicative Content:

- Overview of sexual differentiation, including differentiation of the fetal testes and ovary, and the endocrinology and embryology of sexual differentiation
- The anatomy and physiology of the male reproductive tract
- The anatomy and physiology of the female reproductive tract
- Hormonal control of female reproduction, including the menstrual cycle, follicle growth, autocrine and paracrine factors regulating follicle growth, follicular fluid, ovulation, corpus luteum.
- Hormonal control of male reproduction
- Basic understanding of the regulatory mechanisms associated with human assisted reproductive therapy (ART)
- The roles of ART centre staff: clinicians, scientists, clinical embryologists, nurses, counsellors.
- Principles of and standards for diagnostic semen analysis
- Characteristics of normal and abnormal semen samples
- Semen preparation including different methodologies, diagnostic tests and functional tests

4.0 Timetable

Year 1

Induction National and introductory courses	1 month
Genetics Module 1: Genetics of Learning Disorders	5 months
Rotational module 1:	3 months
Genetics Module 2: Infertility and Disorders of Sexual Differentiation	3 months

Year 2

Genetics Module 2: Infertility and Disorders of Sexual Differentiation	1 month
Rotational Module 2:	3 months
Genetics Module 3: Population Screening	5 months
Rotational module 3:	3 months

Year 3

Genetics Module 4: Cancer	6 months
Research Project	4 – 6 months

5.0 Trainee Portfolio, Assessment and Competency Strategy

You have an **assessment manual**, which must be read in conjunction with this chapter, 'Online assessment and personal development management system' which is available on request from the National School by emailing Genetics.nwd@westmidlands.nhs.uk. In addition to assessment through the academic programme, which will be defined by The University of Nottingham and referred to in the **Course Hand Book**, there will also be continuous assessment across the 3-year training period in the work place, using a series of Directly Observed Procedures (DOPS), Case Based Discussions (CBDs) competence assessment, and Multi Source Feedback (MSF). Examples of CBDs and DOPS can be found at **Appendix 3**.

MSF is an externally applied assessment which provides a sample of attitudes and opinions of colleagues on the technical performance and professional attitude of a trainee. The tool helps to provide data for reflection on performance and gives useful feedback for self evaluation.

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.

Each trainee will have an electronic portfolio in which all of these documents are kept. All assessments will be completed electronically and analysed on a central database.

Any additional documents that are important to a portfolio for example an audit or presentation can be attached electronically to the portfolio.

Each trainee and each assessor will have a personal log in for the online portfolio.

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	Module 4
2 DOPS 1 CBD Competencies	2 DOPS 1 CBD Competencies MSF	2 DOPS 2 CBD Competencies	2 DOPS 2 CBD Competencies MSF

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 2 years and a final 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**.

6.0 Competencies

The competencies form the foundation of the work based training programme and form an important part of the e-portfolio and the trainee's record of competence.

Competencies are transferable across learning outcomes and do not need to be undertaken twice where they are repeated. Reference should be made to the point at which this competency has been completed.

Competencies are cumulative and as such not all competencies have to be completed within the relevant module. All competencies should be completed by the end of the training programme.

This manual provides examples of areas of application or evidence required to demonstrate competence. Trainees are expected to utilise different tools, resource and media within the local laboratory to demonstrate each area of competence.

Some competencies are exit competencies. These are described as such in the recognition that they require longer time and experience to acquire and therefore cannot be limited to one specific module or individual learning outcome. Where possible we have given incremental competencies as a guide.

The competency log book is available online as part of the e-portfolio.

6.1 MODULE 1 Genetics of Learning Disorders

Code	Competency	Reviewer	Date	Comments
M1.1	Is able to describe and summarise the range of genetic testing relevant to learning disabilities.			
M1.2	Demonstrates ability to process a blood sample to obtain chromosome preparations suitable for analysis and appropriate staining and banding techniques relevant to this referral group.			
M1.3	Is competent in chromosome analysis from patients with learning disorders including the use of ISCN.			
M1.4	Interprets results from chromosome analysis using relevant on line databases and search engines.			
M1.5	Knows when to refer for further test – e.g. FISH.			
M1.6	Knows how to set up and analyse samples using a microarray platform suitable for the detection of genome anomalies associated with learning disorders.			
M1.7	Interprets the significance of CNVs using genomic databases.			
M1.8	Demonstrate competence in preparing samples for fragment analysis on a genetic analyser e.g. ABI3130 or equivalent.			
M1.9	Confirm the results of PCR across the <i>FMR1</i> expansion using Southern blot analysis.			
M1.10	Performs a basic analysis and interpretation of fragile X syndrome using results from fluorescent PCR across the <i>FMR1</i> gene expansion in order to detect results within normal, intermediate and expanded ranges, and results which are to be designated as failed analyses.			
M1.11	Knows how to identify samples			

	which require repeating and / or which require Southern blotting.			
M1.12	Knows how to interpret results from Southern blotting for the detection of large expansions and methylation status of alleles in fragile X.			
M1.13	Understands the role of non-routine investigations available to elucidate unusual results further (for example sequencing of <i>FMR1</i> gene).			
M1.14	Describe the basis of manifesting female carriers in X-linked disorders.			
M1.15	Knows how to interpret results from MLPA or bisulphite analysis for PWS/AS syndrome.			
M1.16	Understanding of LIMS to access correct reporting templates.			
M1.17	Demonstrates the use of pedigree analysis for a range of genetic disorders, which can result in learning difficulties.			
M1.18	Know how to access sources of further information for extended testing in learning disability (e.g. use of UKGTN database to identify commissioned tests and where they could be undertaken).			

6.2 Module 2 Infertility and Disorders of Sexual Differentiation

Code	Competency	Reviewer	Date	Comments
M2.1	Is able to describe and summarise the range of genetic testing relevant to infertility and disorders of sexual differentiation.			
M2.2	Demonstrate ability to accurately analyse both normal and abnormal chromosome complements for this group of patients.			
M2.3	Understand and describe the chromosomal abnormalities associated with abnormal sexual differentiation and infertility.			

M2.4	Be able to identify the various Y-chromosome deletions.			
M2.5	Understand and describe the mechanism leading to Y-chromosome microdeletions and the clinical features associated with each class of deletion.			
M2.6	Understand and describe the molecular pathogenesis of the <i>FMR1</i> pre-mutation in relation to premature ovarian failure to both the patient and her family.			
M2.7	Describe the genotype/ phenotype correlations associated with androgen receptor mutations and androgen insensitivity syndrome.			
M2.8	Set up either an ARMS or OLA test to detect common <i>CFTR</i> mutations; be able to describe the advantages and disadvantages of each technique.			
M2.9	Be able to analyse, interpret and report on the most common <i>CFTR</i> mutations.			
M2.10	Understand the implications for other family members of identifying a <i>CFTR</i> mutation in an infertile male.			
M2.11	Understand and describe the molecular pathogenesis of <i>CFTR</i> mutations in relation to bilateral CBAVD.			
M2.12	Demonstrate the ability to interpret data produced during testing and compile accurate reports according to best practice guidelines.			
M2.13	Demonstrate an understanding of sex differentiation disorders in relation to genes such as Androgen Receptor. Understand the basis of skewed X-inactivation.			
M2.14	Demonstrate the ability to describe the implications of the genetic tests (including ethical, legal and social implications) for the effective management of this group of			

	patients.			
M2.15	Understand the counselling issues regarding assisted reproduction in couples with infertility.			
M2.16	Be aware of the counselling issues regarding gender assignment in ambiguous genitalia cases.			
M2.17	Briefly discuss the principles of oogenesis, spermatogenesis and fertilisation with respect to chromosome abnormalities.			
M2.18	Demonstrate awareness of the potential role of Assisted Reproductive Techniques imprinting defects.			

6.3 Module 3 Population Screening

Code	Competency	Reviewer	Date	Comments
M3.1	Describe the basic principles of screening, compliance and choice, cost effectiveness and clinical pathways.			
M3.2	Describe the different approaches to screening including the relationship between the genetic testing and other screening modalities such as USS and IRT.			
M3.3	State and discuss the origin, prevalence and clinical significance of the most commonly encountered chromosome abnormalities seen in chromosome preparations from prenatal samples from screening programs.			
M3.4	Demonstrate an understanding of the utility of non-invasive prenatal diagnosis.			
M3.5	Describe and perform analysis of QF-PCR for the diagnosis of aneuploidy to defined laboratory standards and demonstrate			

	understanding of the technology.			
M3.6	Describe the local policy for reporting results of normal and abnormal QF-PCR analysis.			
M3.7	Recognise and describe the nature and effect of mosaicism, maternal cell contamination, and twin pregnancies in relation to QF-PCR testing.			
M3.8	Demonstrate awareness of the procedures for confirmation of abnormal QF-PCR results.			
M3.9	To perform interphase FISH and demonstrate an understanding of the technology.			
M3.10	To perform and understand long term cell culture techniques necessary for chromosome analysis from prenatal samples.			
M3.11	To perform high resolution chromosome analysis on samples from prenatal samples.			
M3.12	Demonstrate an understanding of the main clinical features of those conditions associated with chromosome abnormalities commonly encountered at prenatal diagnosis.			
M3.13	Demonstrate an understanding of the associations between various abnormal ultrasound scan signs, chromosome abnormalities and genetic disease.			
M3.14	Demonstrate an understanding of a range of testing methods for targeted mutation analysis.			
M3.15	To perform at least one method of targeted mutation analysis using a fast throughput platform.			
M3.16	Demonstrate an ability to interpret the results from a range of tests such as QFPCR, interphase FISH, chromosome analysis and targeted mutation analysis.			
M3.17	To construct accurate, comprehensive and appropriate			

	reports for a range of results associated with aneuploidy testing, chromosome abnormalities and targeted mutation testing.			
M3.18	Compare the ethical, legal and social implications of genetic testing in the context of screening programs with other clinical pathways for example specific diagnostic testing.			
M3.19	Demonstrate an understanding of informed consent relating to screening programs.			

6.4 MODULE 4 Cancer

Code	Competence	Reviewer	Date	Comments
M4.1	To demonstrate the ability to manage the referral and testing process for an inherited cancer gene including the management of predictive testing.			
M4.2	To perform mutation detection in a large gene such as an inherited cancer gene to the required quality standard, including any reflex testing required.			
M4.3	To demonstrate the ability to perform FISH analysis on oncology samples using standard and breakapart/fusion probes.			
M4.4	To demonstrate the ability to perform quantitation of residual disease for example BCR-ABL analysis in CML.			
M4.5	To demonstrate the ability to analyse a range of common chromosomal rearrangements associated with haematological malignancy.			
M4.6	Demonstrate the ability to interpret the relationships between chromosome abnormalities, oncogenes and clinical diagnosis in haematological malignancy.			
M4.7	Demonstrate an understanding of			

	the role of genetic testing in the management of bone marrow transplants.			
M4.8	Demonstrate the ability to produce a report for a range of results of genetic testing in a large cancer gene including diagnostic and predictive testing.			
M4.9	Demonstrate the ability to assess the significance of unclassified variants using appropriate tools such as relevant software.			
M4.10	To calculate an <i>a priori</i> and a <i>posterior</i> risk to an individual in a pedigree of being affected with a disorder.			
M4.11	Demonstrate the ability to produce an integrated report from a range of genetic testing in haematological malignancy.			
M4.12	To understand the benefits to the patients of an integrated report in molecular pathology, for example in MSI and IHC in colorectal cancer.			
M4.13	Demonstrate an understanding of the prognostic value in genetic testing in the management of cancer patients.			
M4.14	Demonstrate an understanding of the utility of guidelines such as NICE and IOG in the management of patients.			
M4.15	Demonstrate an understanding of informed consent relating to predictive testing.			

6.5 MODULE 5 Research Project

Code	Competence	Reviewer	Date	Comments
M5.1	Able to identify an appropriate area for development and its application to clinical practice.			
M5.2	Demonstrates knowledge of scientific experimental design and planning of a robust scientific			

	experiment, including awareness of health and safety, cost implications and ethical issues.			
M5.3	Able to conduct a scientific experiment, including problem solving, relevant data collection and analysis methods and statistical evaluation.			
M5.4	Able to critically evaluate the process and findings of the research and disseminate via a written report.			
M5.5	Shows effective time management skills and workload prioritisation in the context of scientific research and writing.			
M5.6	Able to undertake research within current ethical and governance guidelines and disseminates the research output.			
M5.7	Uses own knowledge and scientific literature to continually review and develop good scientific practice.			
M5.8	Makes optimal use of current best evidence in making decisions and the ability to construct evidence based guidelines and protocols in relation to scientific practise.			
M5.9	Able to perform audits of scientific practice and apply the findings appropriately and complete the audit cycle within current ethical and governance regulations.			

6.6 Generic, Managerial and Leadership Competencies

Code	Competence	Reviewer	Date	Comments
G.1	Able to recognise and accept the responsibilities and role of the scientist in relation to other healthcare professionals.			
G.2	Communicates succinctly and effectively with other professionals as appropriate offering both professional and clinical advice.			
G.3	Able to prioritise and organise			

	duties in order to optimise the work of the department.			
G.4	Able to make appropriate decisions in order to optimise the effectiveness of the scientific team resource.			
G.5	Demonstrates the skills and experience to undertake effective, appropriate leadership and management responsibilities in a genetics laboratory.			
G.6	Demonstrates the ability to teach a range of health professionals and other audiences in a variety of different ways and able to assess the quality of the teaching of self and others.			
G.7	Demonstrates an understanding of the structure of the NHS and the management of local healthcare system.			

Role Descriptor Template for Healthcare Scientists (HCS)

- A Healthcare Scientist (HCS) will have clinical and specialist expertise underpinned by theoretical knowledge and experience and will:
 - Undertake complex scientific and clinical roles, including those working directly with patients
 - Analyse, interpret and compare investigative and clinical options
 - Make judgements, including clinical judgements involving complicated facts or situations that impact on patients
 - Initiate and undertake innovation, improvement and R&D and be involved in education of trainees and other learners in the workplace
-

General Scope in Genetics

Health Care scientists of this grade are responsible for their own work, have a significant role in ensuring the accuracy and quality of the work and work in a range of health care settings

Involved in a medium-term service improvement and innovation including

- Setting of service standards
- Evaluation and implementation of new methods
- Leading development on clinical protocols
- Determine and recommend appropriate testing strategies and consequent clinical protocols
- Clinical audit
- Leading in the identification, evaluation and recommendation for the purchasing and commissioning of new equipment

Patient management and clinical care

- Direct delivery of patient care – contribution to:
- Collection of clinical samples from patients
- Support to specialist clinics for example fetal medicine, genetics oncology, molecular pathology
- Perform, report and interpret a range of investigations undertaken directly with patients within a range of settings
- Advise on prescribing in relation to genomic medicine within all settings
- Provide specialist input to patients across the range of care pathways and health care settings
- Provide appropriate clinical and scientific advice and interpretation of analytical results

- Participate in multidisciplinary meetings for example pathology, haematology, oncology and fetal medicine
- Apply and promote evidence-based practice and use of relevant clinical protocols

Management and Leadership

- Lead teams of staff in development and delivery of defined areas of service
- Lead service improvement and innovation projects in agreed areas of practice
- Performance manage designated section/s of the department to nationally accepted standards and outcomes
- Promote flexible and adaptable leadership response to the demands of the service and to the needs of patients

Communication and working with others

- Communication and interpretation of complex clinical, scientific and technical information to a wide range of people including, clinicians, managers, patients and the public
- Liaise with senior scientists and clinical users of the service on appropriateness of investigations, interventions and tests
- Communicate scientific innovation and service redesign for example by advising primary care and through the use of genomic medicine
- Communicate research and development findings in written and oral formats to internal and external contacts

Education and training

- Clinical, scientific and technical teaching and training of peers, undergraduates, post graduates and other healthcare professionals within relevant areas of practice
- Inform and advise patients and the public on the use of relevant clinical laboratory or scientific services
- Undertake assessment in a variety of settings

Research, Development and Innovation

- Identify research needs and opportunities aimed at improving patient care through genetics
- Management and implementation of R&D projects including translational research for example identification of novel genes and genomic diagnostic testing
- Leading the transformation and application of new technologies for clinical use
- Undertake medium term service development and enhancement including scientific methods development
- Assimilate current research and development data into provision of the most up to date genetic testing for patient care.
- Evaluate, publish developments and innovation

- Contributing to local innovations in health care

Clinical Governance

- Maintain standards for laboratory health and safety procedures
- Comply with quality and governance procedures within the department including risk management and risk mitigation
- Maintain high standards of professional and personal conduct
- Ensure that patient safety and experience and effectiveness of service are maximised
- Perform, analyse and report on relevant clinical audits

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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

Appendix 3

Direct Observation of Practical/Procedural Skills: Template: Genetics STP

Procedure No.	Title				
Clinical Context	Genetics of learning disorders	Infertility and disorders of sexual differentiation	Population Screening	Cancer	Research project

Assessor's Name:	
Assessor's position:	

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 for STP1 completion	Borderline for STP1 completion	Meets expectations for STP1 completion	Above expectations for STP1 completion	CU
1. Understands the clinical context of the procedure including priority setting and testing strategies.					
2. Understands scientific principles of procedure including basic biology underpinning it and an awareness of troubleshooting					
3. Has read, understands and follows the appropriate SOP's, risk and COSHH assessments, and any other relevant H&S documentation including equipment care and maintenance.					
4. Understands and applies the appropriate test validation, IQC,EQA relevant professional/ clinical guidelines					
5. Understands and applies associated IT/bioinformatics					
6. Accurately completes associated documentation					

7. Output meets accepted laboratory/professional standards					
8. Carries out the procedure within appropriate time frame					
9. Is aware of the limitations of the test and sensitivity/specificity					
10. Is able to analyse, interpret and report the results of the procedure and provide appropriate clinical advice					
11. Demonstrates awareness of the limits of responsibility and when to seek advice					
12. Understands the ethical, legal and social implications of the procedure					
13. Consideration of patient/professionalism					
14. Overall ability to perform					

¹ Unable to comment. Please mark this if you have not observed the behaviour

FEEDBACK AND DOCUMENTATION OF LEARNING NEEDS	AGREED ACTION

Outcome	Satisfactory Unsatisfactory	Date of assessment		Time taken for assessment:	
Signature of Assessor		Signature of Trainee		Time taken for feedback:	

Case Based Discussion: Template: Genetics STP

Trainee identification data

Brief description of output and focus of Scenario/Case Discussed:	
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Title of Module:			
Complexity of the Difficulty of scenario:	Low	Average	High

Assessor's Name:			
Assessor's position:	Clinical Scientist Band 7	Clinical Scientist Band 8	Other

Please grade the following areas using the scale below	Below expectations for STP1 completion	Borderline for STP1 completion	Meets expectations for STP1 completion	Above expectations for STP1 completion	Σ
1. Understands the clinical context of the scenario including priority setting and testing strategies.					
2. Understands scientific principles of scenario					
3. Can discuss the relevant procedures involved in the scenario and associated health and safety issues					
4. Understands and applies the appropriate test validation, IQC,EQA relevant professional/ clinical guidelines					
5. Understands and applies associated IT/bioinformatics and other appropriate resources					
6. Is able to interpret and report patient results and provide appropriate clinical advice					
7. Can discuss the significance of patient results within the clinical context of the referral					
8. Understands the ethical, legal and social implications of the scenario					
9. Is aware of the importance of audit and can use this tool effectively					
10. Output meets accepted					

laboratory/professional standards					
11. Demonstrates awareness of the limits of responsibility and when to seek advice					
12. Consideration of patient/professionalism					
13. Overall ability to perform					

¹ Unable to comment. Please mark this if you have not observed the behaviour

FEEDBACK AND DOCUMENTATION OF LEARNING NEEDS	AGREED ACTION

Outcome	Satisfactory Unsatisfactory	Date of assessment		Time taken for assessment:	
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Signature of Assessor		Signature of Trainee		Time taken for feedback:	
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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 (5th edition), Lodish, ed. (2003) Freeman

CLINICAL GENETICS

ABC of Clinical Genetics (3rd edition) Kingston (2002) BMJ Books

Medical Genetics. Young (2005), Oxford Medical Publications

Principles and Practice of Medical Genetics (4th edition) Emery and Rimoin, eds. (2002) Churchill Livingstone

Practical Genetic Counselling (6th 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 (12th edition) Emery, Turnpenny and Ellard eds. (2005) Churchill Livingstone

MOLECULAR GENETICS

Human Molecular Genetics (4th edition) Strachan and Read (Garland 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 (2nd 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 (2nd 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 (2nd 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 2nd Ed 2001

OTHER SUBJECTS

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

Essential Haematology (4th edition). Hoffbrand and Pettit (2001) Blackwell

Essential Immunology (10th 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

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

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