

**HIGHER MEDICAL TRAINING**

**CURRICULUM**

**FOR**

**CLINICAL GENETICS**

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## INTRODUCTION

Clinical Genetics is the specialty concerned with the diagnosis of inherited disorders and birth defects, with the estimation of genetic risks and with genetic counseling of family members. Clinical geneticists generally work in multidisciplinary regional genetic centres, along with scientists, clinical co-workers (genetic counselors and nurses) and academic colleagues.

The specialty of Clinical Genetics is constantly changing and the clinical geneticist must take account of new findings and alter practice accordingly. S/he will also act as an information resource for other medical specialists. Clinical geneticists need a wide range of clinical skills since genetic disorders can affect people of all ages and all body systems. Communication skills are particularly important in explaining complex concepts and test results to families to enable them to choose an appropriate course of action.

The clinical geneticist works closely with clinical scientists who are responsible for cytogenetic, biochemical genetic and molecular genetic laboratories. The clinical geneticist also gives advice to other professionals such as teachers, NHS purchasers and lay organizations. Finally, clinical geneticists should also play a part in public education and public debate about diverse issues that arise from new applications of genetic knowledge.

## Entry Requirements and General Professional Training

Clinical geneticists deal with a very wide range of disorders and, previously, applicants for Higher Medical Training in Clinical Genetics may have varied general professional training. However, the main entry qualifications are MRCP and MRCPCH.

The minimum period of General Professional Training (GPT) is 2 years, but applicants are strongly advised to have had at least 3 years GPT. It is desirable, where possible, that applicants have had post-registration experience in the medical care of both children and adults. Applicants are normally expected to have obtained MRCP or MRCPCH. However, other equivalent qualifications, including MRCPGP, MRCPGP and MRCPGP, will be considered if the applicant has undertaken at least one year of paediatric or adult medicine with 6 months acute unselected take during their period of General Professional Training.

GPT is defined as follows:

A minimum of two years in approved posts with direct involvement in patient care and offering a wide range of experience in a variety of specialties

18 months of the two years must be spent in posts providing experience in the admission and early follow-up of acute emergencies

at least 6 of these 18 months must be spent on a service or services on which emergency take is "unselected"

"unselected take" is defined as acute medical intake encompassing the broad generality of medicine/paediatrics ie not restricted to any single or small group of specialties.

Non-UK graduates without MRCP/MRCPCH who compete for HMT posts must provide evidence of having completed equivalent supervised clinical training to a satisfactory level which has provided equivalent experience and must have obtained a postgraduate qualification equivalent to the MRCP or MRCPCH.

## **Duration and Organisation of Training**

The duration of HMT in Clinical Genetics is four years. The programme to which the trainee is appointed will be based in a regional genetics centre and have named consultant trainers (educational supervisors). One consultant within the same region will act as Programme Director. In each centre, there is a minimum of one consultant per trainee. Representatives of the SAC and JCHMT are responsible for inspection and approval of training programmes.

Trainees who have completed degree courses in genetics may gain exemption from part of the 4-year training programme; up to 6 months credit may be given for an M.Sc. in Clinical Genetics and up to 3 months is awarded for a B.Sc. in Genetics. The SAC may, in individual cases, consider awarding educational credits for other courses or training schemes. However, such exemptions from training may not count if the trainee is already in receipt of educational credit for time spent in relevant research. Twelve months is the total training period that is allowable for any combination of research or degree study. A full 3 years of clinically based, Higher Medical Training is the absolute minimum.

Trainees who wish to undertake part of their clinical genetic HMT overseas must ensure that this is in a recognized Genetics centre with supervision provided, have a personal training programme agreed by the SAC in advance, have independent funding and an agreement from the Postgraduate Dean for out-of-programme training. A minimum of two years clinical training must be undertaken in the base center in the UK, ideally including the final 12 months of training prior to CCST.

A maximum of three months in aggregate of maternity, sickness or other exceptional leave can be counted towards training for CCST at the trainee's request. The trainee is required to confirm their decision about this at the time of organizing the leave period.

It is essential that the trainee should have a thorough basic training in genetics with emphasis on human aspects. The training should embrace clinical, laboratory and theoretical work. In addition, training should include statistics and an introduction to relevant computer applications. Practical experience is necessary, at a basic level, of cytogenetic and molecular genetic techniques.

Depending on the resources of the training department, either a modular approach will be adopted to training in the various aspects of clinical genetics, or a year by year training approach will cover the necessary aspects of the curriculum.

Full participation in genetics clinics, with involvement in every aspect of the background work for each consultation is essential. Specific training in communication and counselling skills is important, and access to group or individual psychological supervision throughout the period of clinical training is strongly encouraged. Evidence-based clinical practice is fostered by journal clubs and other educational activities. Participation in clinical genetics audit is essential. Each trainee must have knowledge of the advances in human biology and the pathological sciences that influence clinical genetics practice. Equally, the applications of genetics in modern health care must be understood within a framework that contains the ethical, social and legal dimensions of the specialty.

## **Research**

A period of supervised research is considered a highly desirable part of HMT in Clinical Genetics. If relevant research has been completed before appointment to the SpR grade, the trainee should inform the SAC just after appointment to the grade so that an accurate estimation of the trainee's CCST date may be made. At the discretion of the SAC, such research training may contribute up to 12 months towards the total duration of HMT. However, trainees may choose not to count time spent in research when estimating the CCST. After entering an approved programme, some trainees wish to spend a longer period in research by stepping aside from clinical training for up to three years.

This is acceptable but only one full year of research will count towards completion of the programme. Trainees can choose whether or not to include 1 year of research time towards CCST and are required to confirm their intention at the time.

Exceptionally, where a trainee has undertaken an extended period of research after obtaining their NTN, some limited, additional training credit may be granted for supervised clinical duties carried out during the period of research, but only if these duties are devoted to general aspects of clinical genetics, and are unrelated to the trainee's research project. Clinical duties that are undertaken in the course of research beyond the initial research year are counted at a rate of no more than 1 session per week, allowing one month's credit to be accumulated per year of research.

## **Appraisal and Assessment**

Appraisals are the formative discussions about a trainee's progress in the specialty that are held between each trainee and his or her educational supervisor. Appraisal should begin within one month of starting a clinical attachment. Appraisals are relatively informal, but should be documented and scheduled in advance. Appraisal should occur at least twice a year. In appraisal, the focus of discussion is the trainee's acquisition of competencies.

Assessment of trainees' progress in the specialty is based upon the standard format of annual review via the RITA process. Particular importance is attached to the Penultimate Year Assessment (PYA). Further details are given in the revised Guide to Specialist Registrar Training (February 1998). As the number of trainees in Clinical Genetics in the UK is relatively small, some regions have organised trainee assessment on a supraregional basis, with neighbouring regions arranging joint assessment panels. Regional Specialty Advisers from the participating regions and a representative of the Postgraduate Dean are involved, and at each PYA there is an SAC representative.

## **Flexible training**

Trainees who are unable to work full-time are entitled to opt for flexible training programmes. EC Directive 93/16/EEC requires that:

*i Part-time training shall meet the same requirements as full-time training, from which it will differ only in the possibility of limiting participation in medical activities to a period of at least half of that provided for full-time trainees;*

*ii The competent authorities shall ensure that the total duration and quality of part-time training of specialists are not less than those of full-time trainees.*

The above provisions must be adhered to. Flexible trainees should undertake a pro rata share of the out of hours duties required of their full-time colleagues in the same programme and at the equivalent stage, where this is applicable.

For details of appointment and funding arrangements for flexible trainees, please see the revised Guide to Specialist Registrar Training (February 1998).

## **Training Record**

A Training Record will be maintained by the trainee. It will be counter-signed as appropriate by the educational supervisors to confirm the satisfactory fulfilment of the required training experience and the acquisition of the competencies that are enumerated in the Specialty Curriculum. It will remain the property of the trainee, and must be produced at the annual assessments.

## 2. AIMS

The training programme in clinical genetics aims to produce clinicians who:

<b>CLINICAL</b>	<b>MANAGEMENT</b>	<b>RESEARCH,TEACHING, LEARNING</b>	<b>AUDIT, CLINICAL GOVERNANCE</b>
<p>Apply knowledge of formal genetics and basic sciences in the diagnosis of genetic disorders</p> <p>Establish diagnoses by history taking, clinical examination and investigations</p> <p>Calculate genetic risks</p> <p>Address all aspects of the healthcare needs of patients and their families</p> <p>Recognise the limitation of their own expertise and know when to seek assistance of colleagues</p>	<p>Work as part of a team</p> <p>Manage time and resources to the benefit of themselves, their patients and colleagues</p> <p>Utilise effectively current methods in information technology</p>	<p>Are effective educators of both patients and colleagues</p> <p>Are able to take responsibility for their own educational needs and the attainment of these needs</p> <p>Plan, conduct and write-up a research project</p> <p>Debate the social, ethical and legal issues that affect the practice of clinical genetics</p> <p>Use skills of life long learning to keep up to date with developments in Clinical Genetics</p>	<p>Develop new clinical practices based on analysis of developments in genetic laboratory diagnostics</p> <p>Carry out clinical audit and act on the results</p> <p>Accept the clinician's role and responsibilities in providing high quality patient care, setting and monitoring standards</p> <p>Participate fully in all Clinical Governance activities</p>

### **3. OBJECTIVES**

#### 3.1

By the end of the educational programme, trainees must have the requisite knowledge, skills and attitudes listed in the curriculum, to diagnose and manage genetic aspects of a wide range of disorders in the following categories, including but not restricted to the conditions specified.

**Cancers - common familial cancers** – breast, ovary, bowel

**Cancers - rare genetic cancer syndromes** – adenomatous polyposis coli, multiple endocrine neoplasia, NF 2, von Hippel Lindau disease.

**Cardiac disorders** – hereditary cardiomyopathies and conduction defects

**Congenital abnormalities** – single and multiple; malformations, deformations and disruptions; fetal and neonatal presentations

**Connective tissue disorders** – Marfan syndrome, Ehlers Danlos syndrome

**Cystic fibrosis**

**Chromosomal disorders** – sporadic and familial; numerical and structural abnormalities

**Deafness** – isolated and syndromic deafness

**Dysmorphic syndromes** – common syndromes as well as some experience with rare disorders

**Fragile X syndrome** – and other X-linked mental retardation syndromes

**Haematological disorders** – haemoglobinopathies, haemophilia, thrombophilia, haemochromatosis

**Huntington disease** – and other adult onset hereditary neurodegenerative disorders

**Inborn errors of metabolism**

**Learning disability** – familial and syndromic causes

**Mitochondrial cytopathies** – mitochondrial myopathies/encephalomyopathies and Leber's optic atrophy

**Multifactorial disorders** – neural tube defects, epilepsies and common adult onset disorders

**Neurogenetic disorders** – Spinal muscular atrophy, spinocerebellar ataxias, hereditary neuropathies, hereditary spastic paraplegia

**Neuromuscular disorders** – myotonic dystrophy, Duchenne, Becker, limb girdl, FSH and Emery Dreifuss muscular dystrophies

**Neurocutaneous syndromes** – neurofibromatosis 1 and tuberous sclerosis

**Ophthalmic genetic disorders** – retinitis pigmentosa

**Pharmacogenetic disorders** – malignant hyperthermia and glucose 6 phosphate dehydrogenase deficiency

**Renal disorders** – adult and infantile polycystic kidney disease

**Skeletal dysplasias** – achondroplasia, osteogenesis imperfecta, spondyloepiphyseal dysplasias

**Teratogens** – alcohol and anticonvulsants

### 3.2

By the end of training, specialist registrars must be able to :

Record and analyse family history data

Obtain the medical history and carry out clinical examination as it relates to genetic diseases

Diagnose genetic disease using clinical evaluation and genetic testing

Choose appropriate investigations and interpret results

Provide accurate information and effective genetic counseling to individuals and families

Write clear summaries of genetic clinic consultations in post-clinic letters to colleagues and patients

Formulate management plans for genetic aspects of genetic/hereditary disorders

Perform risk calculation, including Bayes theorem

Carry out phlebotomy, skin biopsy, hair root extraction and photography

Conduct literature searches and use medical genetic databases

Store and retrieve genetic data in single-disease genetic registers

Work effectively in a team with other colleagues providing genetic services

Liaise appropriately with colleagues from other specialists, including family care workers

Make use lay organizations to support patients and families with genetic diseases

Communicate and explain genetic issues to colleagues and the lay public

Work effectively with colleagues in other disciplines

#### **4. TEACHING, LEARNING AND ASSESSMENT METHODS**

*Trainees will acquire competence through:*

- a. Initial contact with and follow-up of patients
- b. On-the-job training (supervised practice with experienced clinician, scientist and genetic co-worker)
- c. Personal study
- d. Post-graduate education courses, conferences and meetings including Dysmorphology Group Meetings
- e. Tailored clinical experience (for example, in Fetal Medicine Unit, and at specialised clinics such as a Marfan syndrome clinic or genetics-ophthalmology clinic)
- f. Taking part in multidisciplinary team work to plan and deliver service provision (e.g. breast cancer screening, fetal medicine) and individual patient care
- g. Learning research methodology
- h. Carrying out an audit projects
- i. Offering professional advice to and teaching medical and non-medical healthcare trainees, patient support groups and other lay groups
- j. Tailored laboratory experience

*Trainees' competence will be assessed by:*

1. Direct observation (in clinic or from video and audiotape recordings) by a trainer
2. Indirect observation from case note review, case presentations (oral and written), reports from other professionals and colleagues (clinical, scientific, administrative)

*drawing on evidence from Training Record entries*

3. Record of clinical experience (genetic case diagnosis and management)
4. A required number of case reports with reflective commentary
5. A record of training received in research and studies conducted by the trainee
6. An audit report and list of audit activities
7. A list of publications
8. A list of presentations at meetings and conferences
9. A list of courses attended including a counselling course and a management course
10. Reports of satisfactory laboratory attachments

In the following curriculum sections, the above teaching, learning and assessment methods are referred to by letters and numbers in bold italics.

## **5. GENERIC AND SPECIALTY SPECIFIC CURRICULUM SUBJECT MATTER FOR CLINICAL GENETICS**

### **INTRODUCTION**

Defining the objectives of the generic skills of the SpRs in training in any of the medical specialties has relied on two documents; the first is "Good Medical Practice" produced by the GMC; the second is the generic curriculum being developed for the SHOs. We have set out the generic knowledge skills and attitudes (or, more readily assessed, behaviour) that we believe are common to all of the medical specialties. This document should be included at the front of all specialty curricula with amendments appropriate to individual specialty requirements. All SpRs must be able to meet these objectives. No time scale is offered for these competencies but they must all be attested for before completion of training. However failure to achieve satisfactory progress in meeting many of these objectives at an early stage would be cause for concern about the SpRs ability to be adequately trained.

The generic curriculum, amended for Clinical Genetics, has been set out in the following headings (sections 3 – 11 relate to items specific to the Clinical Genetic curriculum, which have been incorporated into the final curriculum document):

1. Good clinical care
  - a) History, Examination, Investigations, Management and Note keeping skills
  - b) Time management and decision making
  - c) Procedures
2. Communication and Counselling skills.
3. Formal and Basic Genetics
4. Common Genetic Referrals
5. Neurogenetics
6. Paediatric Genetics and Dysmorphology
7. Cancer Genetics
8. Prenatal Diagnosis and Fetal Dysmorphology

9. Laboratory Genetics
10. Organisation and Provision of Genetic Services for Populations
11. Joint Specialist Clinics
12. Maintaining Good Medical Practice
  - a) Learning
13. Maintaining trust
  - a) Professional behaviour
  - b) Ethics and Legal Issues
  - c) Patient education and disease prevention
14. Working with Colleagues
15. Team working and Leadership skills
16. Teaching
17. Research
18. Clinical Governance (including risk management , audit and guidelines)
19. Structure and Principles of management
20. Information technology, Computer Assisted Learning and Information Management

## 1. GOOD CLINICAL CARE

### A) History, Examination, Investigations, Management & Notekeeping Skills:

Objective: To be able to establish genetic diagnoses by means of clinical history taking, physical examination and use of appropriate investigations and to provide clinical genetic management for patients and families

<b>Subject</b>	<b>Knowledge</b>	<b>Skills</b>	<b>Attitudes</b>
<b>(i) Pre-clinic preparation</b>	Knowledge of relevant disorder acquired by background reading.	Be able to review medical records and identify information sources including databases and literature searches.	Appreciate the importance of identifying key issues and being prepared to deal with these.
<b>(ii) History</b>	Define the patterns of symptoms found in patients presenting with genetic disease. Recognise reliable and unreliable family history data and identify sources for verification.	Be able to take and analyse a clinical history in a relevant, succinct and logical manner. Be able to overcome difficulties of language, physical and mental impairment. Use interpreters and advocates appropriately. Elicit family history information in a sensitive and understanding manner. Draw complex pedigrees accurately, including consanguinity loops, recording appropriate information.	Show empathy with patients. Appreciate the importance of psychological and social factors of patients and relatives in genetic disease. Attention to detail and accuracy in collecting and checking family history and medical data. Appreciate the confidentiality and ethical issues arising from family history gathering
<b>(iii) Examination</b>	Define the pathophysiological basis of physical signs. Define the clinical signs found in genetic diseases.	Be able to perform a reliable and appropriate examination to elicit relevant signs of genetic disease. Perform examination appropriately in situations involving cultural sensitivity. Understand when additional specialist examination is required.	Respect patients' dignity and confidentiality. Acknowledge cultural issues. Appropriately involve relatives. Appreciate the need for a chaperone.
<b>(iv) Investigations including imaging</b>	Know the predictive value of results of investigations. Define the pathophysiological	Ability to prioritise investigations and interpret the results. Ability to perform investigations competently where relevant. Ability to liaise and discuss investigations with	Understand the importance of working with other health care professionals and team working. Show a willingness to provide explanation to patient as to rationale

	<p>basis of investigations. Define the indications for investigations. Define the risks and benefits of investigations. Know the cost effectiveness of individual investigation.</p>	<p>colleagues and to order them appropriately.</p>	<p>for investigations, and possible unwanted effects. Persistence in seeking diagnoses</p>
<p><b>(v)diagnosis and management</b></p>	<p>Recognise pitfalls in single gene inheritance including variable expressivity and reduced penetrance, somatic and gonadal mosaicism. Be able to formulate differential diagnoses for genetic disorders.</p>	<p>Present genetic information to a patient in a sensitive and understanding manner. Calculate genetic risk in single gene disorders by hand Calculate genetic risk by use of a computer programme Use computerized genetic databases and registers for information retrieval. Present undiagnosed cases to colleagues, including dysmorphology club meetings</p>	<p>Show appropriate attitudes towards patients and their symptoms and be conscious of religious or other philosophical contexts particularly in respect to prenatal diagnosis. Clearly and openly explain management options. Sensitivity in breaking bad news. Appreciate the impact of diagnosing serious genetic conditions on family relationships.</p>
<p><b>(vi) Note keeping, letters etc</b></p>	<p>Use of email, internet and the telephone. Define the structure, function and legal implications of medical records &amp; medico-legal reports. Know the <u>relevance of the data protection</u> pertaining to patient confidentiality</p> <p><i>a,b,f 1,2,3,4</i></p>	<p>Record concisely, accurately, confidentially and legibly the appropriate elements of the history, examination, results of investigations, differential diagnosis and management plan. Date and sign all records. Be able to write outpatient letters to professional colleagues. Be able to write appropriate summary letters to patients and their families. Be able to write appropriate letters in response to complaints.</p> <p><i>a,b 1,2,3</i></p>	<p>Appreciate the importance of timely dictation cost effective use of medical secretaries and the growing use of electronic communication. Be aware of the need for prompt and accurate communication with primary care and other agencies. Show courtesy towards medical secretaries and clerical staff.</p> <p><i>a,b,d,I 1,2,3,4,9</i></p>

## B) Time Management And Decision Making:

Objective: To demonstrate that the trainee has the knowledge, skills and attitudes to manage time and problems effectively.

Subject	Knowledge	Skills	Attitudes
(i) Time management	Know which patients/tasks take priority.	Start with the most important tasks. Work more efficiently as clinical skills develop. Recognise when he/she is falling behind and re-prioritise or call for help.	Have realistic expectations of tasks to be completed by self and others. Willingness to consult and work as part of a team.
(ii) Decision making	Understand clinical priorities for investigation and management. b,d 1,2	Analyse and manage clinical problems. b,d 1,2	Be flexible and willing to change in the light of changing conditions. Be willing to ask for help. b,c 1,2

## C) Procedures:

Objective: to demonstrate proficiency in clinical procedures related to genetics.

Subject	Knowledge	Skills	Attitudes
Phlebotomy	Knowledge of technique	Ability to take blood samples from adults and children, including those with special needs	Understand the stress of the technique and obtain consent
Hair root extracton	Knowledge of technique and indications for use	Ability to collect samples suitable for analysis	Explain procedure appropriately and obtain consent
Skin biopsy	Knowledge of technique and indications for use	Demonstrae ability to obtain samples suitable for analysis	Explain procedure appropriately and obtain consent
Clinical photography	Knowledge of technique Understand importance and confidentiality of photographic records  b 1,2,3	Demonstrate ability to take photographs of sufficient quality for clinical use Use of digital photography and storage of data  b 1,2	Explain the need for clinical photography and obtain consent  b 1,2

## 2. COMMUNICATION SKILLS AND GENETIC COUNSELLING

Objective: Acquire and demonstrate effective communication with patients, relatives and colleagues along with the habit of reflection on personal genetic counseling style and effectiveness. (“counseling” in this context means the transmission of information about genetic disease, risk and reproductive options).

<b>Circumstance</b>	<b>Knowledge</b>	<b>Skills</b>	<b>Attitudes</b>
(i) Within a consultation	<p>Know how to structure the interview to identify the patient’s:</p> <ul style="list-style-type: none"> <li>• concerns/priorities/agenda</li> <li>• expectations</li> <li>• understanding</li> <li>• acceptance</li> </ul>	<p>Listen.</p> <p>Use open questions followed by appropriate closed questions.</p> <p>Avoid jargon and use familiar language.</p> <p>Be able to communicate both verbally and in writing to patients whose first language may not be English in a manner that they understand.</p> <p>Use interpreters appropriately.</p> <p>Communicate effectively with people who have hearing or visual disability.</p> <p>Give clear information and feedback to patients and share information with relatives when appropriate</p> <p>Reassure ‘worried well’ patients, appropriately</p>	<p>Demonstrate an understanding of the need for:</p> <ul style="list-style-type: none"> <li>• involving patients in decisions</li> <li>• offering choices</li> <li>• respecting patients views</li> <li>• dress and appearance to be appropriate to the clinical situation and patient sensibility</li> </ul>
(ii) Breaking bad news	<p>Know how to structure the interview and where it should take place.</p> <p>Be aware of the normal bereavement process and behaviour.</p>	<p>Be able to break bad news in steps appropriate to the understanding of the individual and be able to support distress.</p> <p>Avoid jargon and use familiar language.</p> <p>Encourage questions.</p> <p>Maintain appropriate hope whilst avoiding inappropriate optimism.</p>	<p>Act with empathy, honesty and sensitivity.</p>

<p>(iii) Specific genetic issues</p>	<p>Knowledge of ethnic difference in the incidence of genetic disease.          Understanding of cross-cultural issues including consanguinity and arranged marriages.          Understanding of religious beliefs and attitudes to prenatal diagnosis and assisted reproduction techniques.</p>	<p>Appreciation of factors that influence perception of risk.          Acquisition of “non-directive” counseling skills.          Effective use of co-counsellors.          Communication of genetic information and risk to children and adolescents.          Communication with adults and children with learning disability.          Recognising which patients will benefit from referral on to psychological services.</p>	<p>Appreciate patient and family anxieties, both rational and irrational.          Appreciate that every person is influenced by their own culture and beliefs.          Identify particular ethnic perspectives on genetic diseases.          Appreciate the importance of senior figures / elders in patient education.          Appreciate the importance of genetic nurses and genetic associates.          Cultivate habit of reflection and discussion with colleagues after counseling sessions.          Readiness to alter practice in light of experience and feed-back.</p>
<p>iv) Complaints</p>	<p>Have awareness of the local complaints procedures.          Have an awareness of systems of independent review.</p>	<p>Manage dissatisfied patients / relatives.          Anticipate potential problems.</p>	<p>Act with honesty and sensitivity and promptly.          Be prepared to accept responsibility.</p>
<p>(v) Communication with Colleagues</p>	<p>Know:</p> <ul style="list-style-type: none"> <li>• how to write a problem orientated letter</li> <li>• to communicate with members of the MDT</li> <li>• when to phone a GP</li> <li>• when to phone a patient at home</li> </ul> <p><i>a,b,c,d,e,f 1,2,3,4</i></p>	<p>Use appropriate language.          Select an appropriate communication method.</p> <p><i>a,b,c,e,f 1,2,3,4</i></p>	<p>Be prompt and respond courteously and fairly.</p> <p><i>a,b,c,e,f 1,2,3,4</i></p>

### 3. FORMAL GENETICS AND BASIC SCIENCES

OBJECTIVES	KNOWLEDGE	SKILLS	ATTITUDES
<p>Understand cellular and molecular mechanisms that underpin inheritance in man</p> <p>Identify the social and ethical implications of genetic knowledge</p> <p>Understand patterns of inheritance and undertake risk assessment</p> <p>Have knowledge of emerging genetic technologies and their application (including gene therapy)</p>	<p>The chromosomal basis of heredity (mitosis and meiosis)</p> <p>Mechanisms of origin of numerical and structural chromosome abnormalities</p> <p>Behaviour of structural chromosome abnormalities at meiosis</p> <p>The chemical structure of DNA and replication</p> <p>Central dogma of cell biology: transcription and translation</p> <p>Modes of inheritance (Mendelian and non Mendelian)</p> <p>Risk calculations including combinatorial probability and Bayes Theorem</p> <p>The clinical embryology and molecular mechanisms of human malformation syndromes</p> <p>Principles of teratogenesis and pregnancy associated risks</p>	<p>Recognition of different inheritance patterns in pedigrees</p> <p>Pedigree-based calculation of segregation ratios for structural chromosome abnormalities</p> <p>Empiric risk calculations (occurrence and recurrence risks)</p> <p>Perform Bayesian risk calculations including linkage-based risk calculations</p> <p>Calculate gene frequencies - the Hardy-Weinberg equilibrium and chi square tests of departure</p> <p>Apply knowledge to interpret results of chromosome and molecular genetic analysis.</p> <p><i>b,c,d 1,2,3,4</i></p>	<p>Identification and critical evaluation of information</p> <p>Commitment to lifelong self directed learning</p> <p>Use primary sources of data</p> <p>Appreciation the impact of genetic disorders on individuals and families</p> <p>Appreciate potential benefits and harm of new genetic technologies</p> <p>Appreciate public concerns about the application of new genetic technologies</p> <p><i>c,d,g,h 1,2,4-9</i></p>

	Mechanisms of mutagenesis and estimation of mutation rates  History of genetics  <i>c,d,i,j 1,2,3,4</i>		
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#### 4. COMMON GENETIC REFERRALS

OBJECTIVES	KNOWLEDGE	SKILLS	ATTITUDES
<p>To provide the trainee with the skills and knowledge to be able to carry out specialist diagnosis, assessment and genetic counselling for the conditions listed in Section 3</p>	<p>The genetic basis and clinical features of common genetic conditions (see 3.1)</p> <p>The medical and surgical complications of common genetic conditions and indications for referral for specialist opinion</p> <p>Whether molecular/cytogenetic testing is available and its application to diagnosis, predictive testing, carrier testing and prenatal diagnosis</p> <p>Application and limitations of current tests</p> <p>Knowledge of current clinical treatments for 'core' conditions and gene therapy trials</p> <p><i>a,b,e,f 1,2,3</i></p>	<p>Be able to take a relevant history, perform an appropriate examination and formulate clinical diagnoses</p> <p>Be able to assess patients and families affected by genetic conditions</p> <p>Judge when it is necessary to sustain supportive relationships with patients with chronic disease</p> <p>Work in a team to develop and implement long term management utilising evidence based medicine and care pathways</p> <p>Be able to discuss reproductive options (AID, ICSI, IVF, pre-implantation diagnosis) with the patient and their partner in a sensitive manner</p> <p>Be able to discuss and formulate management plans with individuals/families</p> <p><i>a,b,f 1,2,3</i></p>	<p>Value the contribution and role of other specialists</p> <p>Appreciate role of patient education, e.g. in type 1 neurofibromatosis</p> <p>Appreciate the role of the general practitioner in management of chronic disease</p> <p>Appreciate the role of support groups and be willing to provide appropriate information</p> <p>Apply good clinical care and counselling skills (see sections 1 and 2)</p> <p><i>a,b,f,i 1,2</i></p>

## 5. NEUROGENETICS

OBJECTIVES	KNOWLEDGE	SKILLS	ATTITUDES
To provide the trainee with the skills and knowledge to recognise genetic causes of central and peripheral nervous system dysfunction	Genetic aspects and clinical presentation of trinucleotide repeat disorders	Recognise family history data that suggest familial neurological disease	Appreciation of family stresses caused by risk or eventuality of neurodegeneration
	Classification and molecular basis of common genetic neuromuscular disorders	Verify diagnoses from old hospital records	Appreciate social problems encountered by adults with mild/moderate learning disability
	Basic neuropathology and differential diagnosis of hereditary dementias	Be able to confirm clinical signs in affected individuals	Appreciate issues involved in predictive testing
	Mitochondrial diseases – clinical, biochemical and genetic features	Be able to draw up a differential diagnosis and institute appropriate genetic testing	<i>e,f</i> 1,2
	Genetic causes of mental retardation (static and progressive)	Assessment of symptoms and signs in patients at risk of adult-onset neurogenetic disease	
	Genetic contribution to autism and autistic spectrum disorders	Application of protocols for pre-symptomatic diagnosis of Huntington's disease and other neurodegenerative disorders	
	Genetic contribution to psychiatric disease in adults	Make timely, appropriate referrals to other specialists such as neurologists, psychologists, psychiatrists, speech therapists	
<i>a,b,e</i> 1,2,3	<i>a,b,e,f</i> 1,2,3		

## 6. PAEDIATRIC GENETICS AND DYSMORPHOLOGY

OBJECTIVES	KNOWLEDGE	SKILLS	ATTITUDE
To provide the trainee with the skills and knowledge to make syndrome diagnosis in children	<p>Identify normal developmental milestones and diagnose delayed development</p> <p>Explain morphogenesis in terms of deformation, malformation, disruption and dysplasia</p> <p>Utilise journals and databases used in syndrome identification</p> <p>Have knowledge of common and rarer dysmorphic syndromes</p> <p><i>a,b,c,d,e 1,2,3,4</i></p>	<p>Be able to take a relevant history, and perform an appropriate examination, obtain illustrative photographs</p> <p>Have a rational approach to investigation of children with delayed development and/or dysmorphic syndromes</p> <p>Formulate differential diagnoses of unknown syndromes</p> <p>Cultivate critical assessment of database information and case reports to identify uncertainty and subjectivity in syndrome diagnosis</p> <p>Be able to provide a diagnostic service within a multidisciplinary clinical team</p> <p>In malformation syndromes, refer patients appropriately to specialist medical and surgical services</p> <p>Present and discuss cases with colleagues</p> <p><i>a,b,e,f 1,2,3</i></p>	<p>Recognise importance of clinical judgement, timing, and tact when diagnosing and informing parents of an infant with serious malformation or handicap</p> <p>Appreciate the emotional reactions of parents following early diagnosis of syndrome or recognition of developmental delay</p> <p>Appreciate the adverse reaction families may experience following retraction of a previous diagnosis</p> <p>Appreciate the importance of attending Dysmorphology Group Meetings</p> <p><i>a,b,d 1,2,3,8,9</i></p>

## 7. CANCER GENETICS

<b>OBJECTIVES</b>	<b>KNOWLEDGE</b>	<b>SKILLS</b>	<b>ATTITUDES</b>
<p>Trainee is able to diagnose rare cancer syndromes and recognise when common cancers are likely to have a single gene basis</p> <p>The trainee can recommend targeted screening in individuals who are identified as having increased risk</p> <p>Trainee can coordinate appropriate molecular genetic testing</p>	<p>The genetic and environmental factors that affect risk of developing cancer</p> <p>Current recommendations concerning tumour surveillance in cancer -prone families</p> <p>Knowledge of clinical features of genetic cancer syndromes</p> <p>Genetic mechanisms in neoplasia: Knudson's two-hit hypothesis</p> <p>Knowledge of molecular basis of cancer genetic syndromes</p> <p><i>a,b,c,d 1,2,9</i></p>	<p>Be able to take a relevant history, perform an appropriate examination and undertake risk estimation using a variety of methods</p> <p>Use of cancer registers and other sources to verify diagnoses</p> <p>Use disease registers (e.g. von Hippel Lindau disease) to support follow-up of affected and at-risk patients</p> <p>Assessment of screening protocols for at-risk relatives</p> <p>Identify at-risk patients and relatives who are eligible to participate in trials of cancer prevention strategies</p> <p><i>a,b,d,e 1,2,6</i></p>	<p>Demonstrate awareness of the roles primary care and genetic associates play in assessing families where relatives are at risk of developing cancer</p> <p>Inform patients about lifestyle factors that affect cancer risk</p> <p>Support general practitioners with the long-term management of selected patients with familial cancer syndromes</p> <p>Liase with other specialists as appropriate e.g. for advice about prophylactic mastectomy and work as a member of a multidisciplinary team</p> <p>Understand the impact of cancer risk on individuals and families</p> <p><i>a,b,f, 1,2,3</i></p>

## 8. PRENATAL DIAGNOSIS AND FETAL DYSMORPHOLOGY

OBJECTIVES	KNOWLEDGE	SKILLS	ATTITUDES
To provide the trainee with the skills and knowledge to undertake genetic assessment of actual and potential problems in the fetus, and provide parents with advice about prognosis and inheritance	<p>Through attendances at fetal post-mortem examinations, know the process and limitations of clinical and laboratory diagnostic procedures</p> <p>Have knowledge of RCPATH guidelines on retention and storage of fetal tissues</p> <p>Know the natural history of prenatally diagnosed conditions including autosomal and sex chromosome aneuploidy syndromes</p> <p>Knowledge of the law pertaining to termination of pregnancy for fetal abnormality</p> <p><i>c,e 1,3</i></p>	<p>Interpret family history data and trace old medical records</p> <p>Perform post-mortem clinical analysis of the fetus (examination, measurements, photography, radiology, tissue sampling and storage for diagnostic studies)</p> <p>Use syndrome databases in syndrome diagnosis</p> <p>Provide genetic advice for women who may undergo prenatal diagnosis</p> <p>Assess clinical significance of chromosome, DNA, and fetal imaging studies in the context of fetal abnormality or risk thereof</p> <p>Formulate differential diagnoses and assess prognosis in collaboration with the fetal medicine team</p> <p>Perform risk-assessment when pregnancies are exposed to hazards such as congenital infections, alcohol, ionising irradiation or drugs</p> <p>Sensitive disclosure of abnormal test results or diagnoses in the antenatal period</p> <p><i>a-f 1-4</i></p>	<p>Appreciate the different perspectives on advantages and disadvantages of prenatal diagnosis in each situation</p> <p>Non-judgmental appreciation of the ethical and religious dimensions to prenatal diagnosis</p> <p>Awareness of the adverse psychological effects of termination of pregnancy for fetal abnormality</p> <p><i>i 1,2</i></p>

## 9. LABORATORY GENETICS

OBJECTIVE	KNOWLEDGE	SKILLS	ATTITUDES
<p>The trainee acquires skills and knowledge to interpret genetic laboratory results within a clinical setting, by completing an attachment in the genetic laboratories</p>	<p>Techniques for conventional chromosome analysis in different tissues</p> <p>Laboratory techniques and application of new cytogenetic tests e.g. FISH/CGH</p> <p>Use of ISCN nomenclature</p> <p>Molecular genetic techniques in common usage- (DNA extraction, Southern Blotting, PCR, DNA sequencing)</p> <p>Application of DNA-based testing for gene mapping, linkage and mutation detection.</p> <p>Potential application of new DNA technologies</p> <p>Sensitivity and specificity of laboratory tests</p> <p>Use of DNA and molecular cytogenetic methods in preimplantation diagnosis</p> <p>Investigative approach to biochemical diagnosis of inborn</p>	<p>Interpretation of clinical consequences of abnormal karyotypes, enzyme deficiencies and molecular test results</p> <p>Liaise with molecular and cytogenetics scientists in analysis of test results</p> <p>Provide advice to laboratory on the wording of reports to referring clinicians</p> <p>Genetic risk calculation based on laboratory test results (e.g. MLINK, Bayesian analysis).</p> <p><i>e,f,j 1,2,5</i></p>	<p>Awareness of the importance of informed consent that arise in relation to storage of DNA samples and cell lines</p> <p>Willingness to liaise with colleagues to interpret laboratory results</p> <p><i>f,j 1,2</i></p>

	<p>errors of metabolism (via experience gained at metabolic disease clinics)</p> <p>The operation of local and national antenatal and newborn, genetic disease screening programmes</p> <p><i>d,e,f,j, 1,2,8,9</i></p>		
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## 10. ORGANISATION AND PROVISION OF GENETICS SERVICES FOR POPULATIONS

OBJECTIVES	KNOWLEDGE	SKILLS	ATTITUDES
<p>To identify practical, legal and ethical issues arising from operation of genetic registers</p> <p>To know the criteria against which screening programmes for genetic diseases and susceptibilities are judged</p>	<p>The genetic characteristics in different populations, mutant gene frequencies and disease prevalences</p> <p>The factors that influence decisions to instigate programmes of population screening for genetic diseases</p> <p>Define sensitivity, specificity, and predictive values of screening tests</p> <p>Knowledge of current screening programmes</p> <p>Knowledge of appropriate population-based registers.</p> <p><i>c,j,g 5,9</i></p>	<p>Team-working with database managers, genetic associates and nurse specialists in:</p> <p>- 'cascade screening' and provision of genetic services for extended families with common single gene disorders (cystic fibrosis, Xp21 muscular dystrophy, fragile X syndrome, Huntington's disease)</p> <p>- family based screening for individuals at high risk of developing cancer</p> <p>- contribute to the maintenance of departmental genetic register systems</p> <p>Be able to explain the benefits and consequences of screening programmes</p> <p><i>a,b,f,g 1,2,3,8,9</i></p>	<p>Appreciate ethical and social dimensions to population screening</p> <p>Understand the central role of patient education</p> <p>Appreciate the value of specialised clinics (breast clinics, lipid and cardiovascular risk factor clinics)</p> <p>Encourage patients to adopt a healthier lifestyle with specific emphasis on risk factor avoidance and promotion of behaviours that reduce risk of developing disease</p> <p><i>e,f, 1,2,9</i></p>

## 11. JOINT SPECIALIST CLINICS

OBJECTIVES	KNOWLEDGE	SKILLS	ATTITUDES
To equip the trainee with skills and knowledge to provide genetic advice within multidisciplinary clinic settings	<p>Genetic contribution to multidisciplinary clinics held with other specialists:</p> <p>Examples of such clinics include:</p> <p>Child development Vision Hearing Endocrine Skeletal dysplasia Neurological Cranio-facial malformation Tumour surveillance</p> <p><i>e, f 1,2</i></p>	<p>Team working skills</p> <p>Develop a special interest clinic</p> <p>Develop skills and liaisons needed to nurture new services, even in settings such as health centres or child development centres, outside of the genetics department</p> <p><i>f, 1,2</i></p>	<p>Demonstrate appreciation of the role of other professionals and willingness to be flexible in responding to the needs of colleagues and patients</p> <p><i>f, 12</i></p>

## 12. MAINTAINING GOOD MEDICAL PRACTICE

### A) LEARNING:

**Objective: To inculcate the habit of life long learning**

SUBJECT	KNOWLEDGE	SKILLS	ATTITUDES
<b>Life long learning</b>	<p>Define continuing professional development.</p> <p><i>c 1,2,3</i></p>	<p>Recognise and use learning opportunities.</p> <p>Use the potential of study leave to keep oneself up to date.</p> <p><i>b,c 1,2,3</i></p>	<p>Be:</p> <ul style="list-style-type: none"> <li>• self motivated</li> <li>• eager to learn,</li> </ul> <p>Show:</p> <ul style="list-style-type: none"> <li>• Willingness to learn from colleagues.</li> <li>• willingness to accept criticism.</li> </ul> <p><i>b,c 1,2</i></p>

### 13. MAINTAINING TRUST

#### A) PROFESSIONAL BEHAVIOUR:

**Objective: To ensure that the trainee has the knowledge, skills and attitudes to act in a professional manner at all times.**

<b>SUBJECT</b>	<b>KNOWLEDGE</b>	<b>SKILLS</b>	<b>ATTITUDES</b>
<b>(i) Continuity of care</b>	Understand the relevance of continuity of care.	Ensure satisfactory completion of reasonable tasks at the end of the shift/day with appropriate handover Documentation of/for handover. Make adequate arrangements to cover leave.	Recognise the importance of: <ul style="list-style-type: none"> <li>• punctuality</li> <li>• attention to detail.</li> </ul>
<b>(ii) Doctor-patient relationship</b>	Understand all aspects of a professional relationship. Establish the limiting boundaries surrounding the consultation. Deal with challenging behaviour in patients which transgress those boundaries, eg aggression, violence, racism and sexual harassment.	Help the patient appreciate the importance of cooperation between patient and doctor. Develop a relationship that facilitates solutions to patient's problems. Deal appropriately with behaviour falling outside the boundary of the agreed doctor patient relationship in patients, e.g. aggression, violence, sexual harassment.	Adopt a non-discriminatory attitude to all patients and recognise their needs as individuals. Seek to identify the health care belief of the patient. Acknowledge patient rights to accept or reject advice. Secure equity of access to health care resources for minority groups. Act with compassion at all times
<b>(iii) Recognises own limitations</b>	Know the extent of one's own limitations and know when to ask for advice.	Reflection on individual practice	Be willing to consult and to admit mistakes.
<b>(iv) Stress</b>	Know the effects of stress Have knowledge of support facilities for doctors.	Develop appropriate coping mechanisms for stress and ability to seek help if appropriate.	Recognise the manifestations of stress on self & others.

<p><b>(v) Relevance of outside bodies</b></p>	<p>Have an understanding of the relevance to professional life of:</p> <ul style="list-style-type: none"> <li>• The Royal Colleges</li> <li>• GMC</li> <li>• Postgraduate Dean</li> <li>• Defence unions</li> <li>• BMA</li> </ul> <p>BSHG / CGS Human Genetic Commission GENGAC Patient representation groups Other organization relevant to genetics</p>	<p>Recognise situations when appropriate to involve these bodies/individuals.</p>	<p>Be open to constructive criticism. Accept professional regulation. Respect the views of patient representation groups.</p>
<p><b>(vi) Personal health</b></p>	<p>Know about occupational health services. Know about one's responsibilities to the public. Know not to treat oneself or one's family.</p> <p><i>b,c 1,2</i></p>	<p>Recognise when personal health takes priority over work pressures and to be able to take the necessary time off.</p> <p><i>b,c 1,2</i></p>	<p>Recognise personal health as an important issue.</p> <p><i>b,c 1,2</i></p>

## **B) ETHICS AND LEGAL ISSUES:**

**Objective: To ensure the trainee has the knowledge and skills to deal appropriately with ethical and legal issues that Arise during the management of patients with genetic disorders.**

<b>SUBJECT</b>	<b>KNOWLEDGE</b>	<b>SKILLS</b>	<b>ATTITUDES</b>
(i) Informed consent	Know the process for gaining informed consent Understand process of consent for tissue/sample storage and use. How to gain consent for a research project	Give appropriate information in a manner patients understand and be able to gain informed consent from patients  Appropriate use of written material	Consider the patient's needs as an individual
(ii) Confidentiality	Be aware of relevant strategies to ensure confidentiality. Be aware of situations when confidentiality might be broken	Use and share all information appropriately  Avoid discussing one patient in front of another  Be prepared to seek patients wishes before disclosing information	Respect the right to confidentiality.
(iii) Legal issues relating to: Criminal matters	Know where to seek advice relating to responsibilities in serious criminal matters.	Be able to obtain suitable evidence or know whom to consult if in doubt.	Recognise the importance of legal issues in medical practice and always be ready to seek advice.

<p>iv) Ethical issues relating particularly to clinical genetics</p>	<p>Be aware of professional guidelines published by the GMC, BSHG, CGS and other bodies related to clinical genetics</p> <p>b 1,2,4</p>	<p>Avoid giving “directive” genetic advice          Be able to communicate ethical issues with patients, colleagues and the public, surrounding:          Confidentiality          Informed consent          Predictive genetic testing          Genetic testing of children          Population screening for genetic disease          Assisted reproduction          Prenatal/preimplantation diagnosis          Late termination of pregnancy</p> <p>b,c 1,2,4,9</p>	<p>Appreciate the diversity of public opinion on ethical and moral aspects of the practice of clinical genetics.          Respect opinions of patients.          Respect the opinion of colleagues.          Be prepared to discuss difficult cases with experienced colleagues and take advice.          Be willing to refer on to a colleague if conflict exists between personal values and those of the patient.</p> <p>b,c 1,2</p>
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### **C) PATIENT EDUCATION AND DISEASE PREVENTION:**

**Objective: To ensure that the trainee has the knowledge, skills and attitudes to be able to educate patients effectively**

**About genetic disease.**

<b>SUBJECT</b>	<b>KNOWLEDGE</b>	<b>SKILLS</b>	<b>ATTITUDES</b>
(i) Educating patients about: <ul style="list-style-type: none"> <li>• disease</li> <li>• investigations management</li> </ul>	Know disease course and manifestations. Know investigation procedures including possible alternatives / choices. Be aware of management strategies for genetic disease.	Give information to patients clearly in a manner that they can understand including written information. Encourage questions. Discuss management plans and follow up arrangements	Consider involving patients in developing mutually acceptable investigation plans. Encourage patients to access: <ul style="list-style-type: none"> <li>• further information</li> <li>• patient support groups</li> </ul>
(ii) Environmental & lifestyle risk factors	Understand the risk factors that may influence certain genetic diseases, including; Life style Smoking Alcohol Medication	Advise on lifestyle changes. Advise on teratogenic potential of medication. Involve other health care workers as appropriate.	Do not display prejudice
(vi) Epidemiology & screening	Know the methods of data collection and their limitations. Know principles of 1 <sup>o</sup> & 2 <sup>o</sup> prevention & screening.  b,c,d,e,f,I 1,2,3,4	Assess an individual patient's risk factors. Encourage participation in appropriate disease prevention or screening programmes.  b,c,e,f 1,2,4	Consider the: <ul style="list-style-type: none"> <li>• positive &amp; negative aspects of prevention</li> <li>• importance of patient confidentiality</li> </ul> Respect patient choice.  B,c 1,2,4

## 14. WORKING WITH COLLEAGUES:

**Objective: To demonstrate good working relationships with Colleagues**

<b>SUBJECT</b>	<b>KNOWLEDGE</b>	<b>SKILLS</b>	<b>ATTITUDES</b>
(i) Interactions between: <ul style="list-style-type: none"> <li>• hospital &amp; GP</li> <li>• hospital &amp; other agencies e.g. social services</li> <li>• medical and surgical specialties</li> </ul>	Know the roles and responsibilities of team members. Know how a team works effectively. Know the roles of other clinical specialties and their limitations. Know the role of multidisciplinary management in genetic disorders.	Show leadership, delegate and supervise safely Be able to communicate effectively. Handover safely. Seek advice if unsure. Recognise when input from another specialty is required for individual patients. Be able to work effectively with GPs, other medical and surgical specialists and other health care professionals.	Show respect for others opinions. Be conscientious and work co-operatively. Respect colleagues, including non medical professionals, and recognise good advice. Recognise own limitations.
	<i>b,c 1,2</i>	<i>b,f 1,2</i>	<i>b 1,2</i>

## 15. TEAM WORKING & LEADERSHIP SKILLS

**Objective: To demonstrate the ability to work in clinical teams and to have the necessary leadership skills**

<b>SUBJECT</b>	<b>KNOWLEDGE</b>	<b>SKILLS</b>	<b>ATTITUDES</b>
Clinical teams.  Respect others opinion  Effective leadership skills	Roles & responsibilities of team members. How a team works. Ensuring colleagues understand the individual roles and responsibilities of each team member. Own professional status and specialty A knowledge of the field. The capacity to perceive the need for action and initiate that action  <i>b,c,d 1,2</i>	Respect skills and contribution of colleagues to be conscientious and work constructively. Respect for others opinion. To recognise your own limitations Objective setting; Lateral thinking; Planning; Motivating; Organising; Setting example; Negotiation skills.  <i>b,c 1,2</i>	Recognise own limitations. Enthusiasm; integrity; courage of convictions; imagination; determination; energy; and professional credibility.  <i>b,c 1,2</i>

## 16. TEACHING AND EDUCATIONAL SUPERVISION:

**Objective: To demonstrate the knowledge, skills and attitudes to provide appropriate teaching, learning and assessment opportunities in clinical genetics for varied groups (medical, other health professional and lay groups)**

<b>SUBJECT</b>	<b>KNOWLEDGE</b>	<b>SKILLS</b>	<b>ATTITUDES</b>
(i) To have the skills, attitudes and practices of a competent teacher (through participation in a recognized course for medical educators)	The goals and objectives of undergraduate medical education as set out by the GMC. Identify adult learning principles. Identify learner needs. Identify learning styles. Structure teaching activities for large audiences, small groups and clinic based teaching. Principles of evaluation.	Facilitate learning process. Identify learning outcomes. Construct educational objectives. Design and deliver an effective teaching event. Communicate effectively with the learners. Use effective questioning techniques. Teach large and small groups effectively. Select and use appropriate teaching resources. Give constructive effective feedback. Evaluate programmes and events Use different media for teaching that are appropriate to the teaching setting. Be able to chair an educational event.	Demonstrate a willingness, enthusiasm and commitment to teach. Show respect for the learner. Demonstrate a professional attitude towards teaching. Demonstrate a learner centred approach to teaching. Seek feedback and demonstrate a willingness to change methods in response to constructive feedback.
(ii) Assessment	Know the principles of assessment Know different assessment methods Define formative and summative assessment	Use appropriate assessment methods Give constructive, effective feedback	Be honest and objective when assessing performance.
(iii) Appraisal	Know the principles of appraisal Know the structure of the appraisal interview	Participate in effective appraisal	Show respect for those participating in appraisal.

## 17. RESEARCH [WHERE UNDERTAKEN]

Trainees are encouraged to undertake a period of full time research and have a good knowledge of research methodology. There should be active involvement with research projects throughout the training period.

SUBJECT	KNOWLEDGE	SKILLS	ATTITUDES
To be able to plan and analyze a research project.	<p>Be able to set up a hypothesis and test it.</p> <p>Know how to design a research study.</p> <p>Know how to use appropriate statistical methods.</p> <p>Know the principles of research ethics and the role of ethical committees (LREC, MREC).</p> <p>Know how to write a scientific paper.</p> <p>How to identify sources of research funding.</p> <p><i>c,d,g</i> 5</p>	<p>Undertake systematic critical review of scientific literature.</p> <p>Ability to frame questions to be answered by a research project.</p> <p>Develop protocols and methods for research.</p> <p>Obtain ethical committee approval for a research proposal.</p> <p>Participate in collaborative research with clinical/scientific colleagues.</p> <p>Be able to use databases.</p> <p>Be able to accurately analyse data.</p> <p>Write and submit a case report or scientific paper.</p> <p>Have good written and verbal presentation skills.</p> <p><i>c,d,g,j</i> 1,5,7,8</p>	<p>Demonstrate curiosity and a critical spirit of enquiry.</p> <p>Demonstrate the persistence needed to follow a project from inception to completion.</p> <p>Ensure patient confidentiality.</p> <p>Demonstrate knowledge of the importance of ethical approval and patient consent for clinical research.</p> <p>Humility and the acknowledgement of the contribution of others.</p> <p><i>a,b,c,d,g</i> 2,5,7,8</p>

## 18. CLINICAL GOVERNANCE

**Objective: Demonstrate an understanding of the context, the meaning and the implementation of Clinical Governance.**

SUBJECT	KNOWLEDGE	SKILLS	ATTITUDES
<p>(i) The organisational framework for Clinical Governance at local, health authority and national levels.</p> <p>Understanding of the benefits a patient might reasonably expect from Clinical Governance.</p> <p>Creating an environment where mistakes and mismanagement of patients can be openly discussed and learned from</p>	<p>Define the important aspects of Clinical Governance.</p> <ul style="list-style-type: none"> <li>• Medical and clinical audit.</li> <li>• Research and Development.</li> <li>• Integrated care pathways.</li> <li>• Evidenced based practice.</li> <li>• Clinical effectiveness.</li> <li>• Clinical risk systems.</li> <li>• To define the procedures and the effective action when things go wrong in own practice or that of others.</li> <li>• Complaints procedures.</li> </ul> <p>Quality assurance schemes in genetic laboratory practice</p>	<p>Be an active partaker in clinical governance.</p> <p>Be able to undertake medical and clinical audit. Be actively involved in audit cycles.</p> <p>Be active in research and development.</p> <p>Critically appraise medical data research.</p> <p>Practice evidence based medicine.</p> <p>Aim for clinical effectiveness (best practice) at all times.</p> <p>Educate self, colleagues and other health care professionals.</p> <p>Be able to handle and deal with complaints in a focused and constructive manner.</p> <p>Learn from complaints.</p> <p>Develop and institute clinical guidelines and integrated care pathways. Be aware of advantages and disadvantages of guidelines.</p> <p>Report and investigate critical incidents.</p> <p>Regular review of adverse events and modify practice accordingly.</p> <p>Take appropriate action if you suspect you or a colleague may not be fit to practice.</p>	<p>Make the care of your patient your first concern.</p> <p>Respect patients privacy, dignity and confidentiality.</p> <p>Be prepared to learn from mistakes, errors and complaints.</p> <p>Recognise the importance of team work.</p> <p>Share best practice with others.</p> <p>Willingness to cultivate a questioning approach to current practice of clinical genetics and motivation to make improvements.</p>

(ii) Risk management	<p>Knowledge of such matters as H&amp;S policy, policies on needlestick injuries, note keeping, communications and staffing numbers.</p> <p>Knowledge of risk assessment, perception and relative risk</p> <p>Know the complications and side effects of treatments.</p>	<p>Confidently and authoritatively discuss risks with patients and to obtain informed consent.</p> <p>Able to balance risks and benefits with patients.</p>	<p>Willingness to respect and accept patients views and choices</p> <p>Willingness to be truthful and to admit error to patients, relatives and colleagues.</p>
(iii) Evidence	<p>Know &amp; understand:</p> <ul style="list-style-type: none"> <li>• the principles of evidence based medicine</li> <li>• the types of evidence</li> </ul>	<p>Able to critically appraise evidence.</p> <p>Ability to be competent in the use of databases, libraries and the internet.</p> <p>Able to discuss the relevance of evidence with individual patients</p>	<p>Display a keenness to use evidence in the support of patient care and own decisions therein.</p>
(iv) Audit	<p>Know &amp; understand:</p> <ul style="list-style-type: none"> <li>• the audit cycle</li> <li>• data sources</li> <li>• data confidentiality</li> </ul>	<p>Involvement in on-going audit.</p> <p>Undertake at least one audit project</p>	<p>Consider the relevance of audit to:</p> <ul style="list-style-type: none"> <li>• benefit patient care</li> <li>• clinical governance</li> </ul>
(v) Guidelines	<p>Know the advantages and disadvantages of guidelines</p> <p>Methods of determining best practice</p> <p><i>b,c,d,f,h 4,6,9</i></p>	<p>Ability to utilise guidelines</p> <p>Be involved in guideline generation, evaluation, review and updating.</p> <p><i>b,c,f,I 2,4,6</i></p>	<p>Show regard for individual patient needs when using guidelines</p> <p>Willingness to use guidelines as appropriate</p> <p><i>a,b,I 1,2,4,9</i></p>

## 19. STRUCTURE OF THE NHS AND THE PRINCIPLES OF MANAGEMENT

**Objective: To display a knowledge of the structure and organisation of the NHS nationally and locally.**

SUBJECT	KNOWLEDGE	SKILLS	ATTITUDES
<p><b>Structure of the NHS and the principles of management</b></p>	<p>Know the structure of the NHS, primary care groups, Trusts and Hospital Trusts. Know the local Trusts structure including Chief Executives, Medical Directors, Clinical Directors and others. Know the role of postgraduate deaneries, specialist societies, the royal colleges and the general medical council.</p> <p>Know finance issues in general in the Health Service, especially budgetary management.</p> <p>Know the appointments procedures and the importance of equal opportunities.</p> <p>Know of Central Government health regulatory agencies (eg NICE, CHI)</p> <p><i>c,d</i> 9</p>	<p>Develop skills in managing change and managing people.</p> <p>Develop leadership skills to play a leading role in developing local genetic services.</p> <p>Develop interviewing techniques and those required for performance reviews.</p> <p>Be able to build a business plan.</p> <p><i>c,d</i> 9</p>	<p>Show an awareness of equity in health care access and delivery.</p> <p>Demonstrate an understanding of the importance of a health service for the population.</p> <p>Show respect for others, ensuring equal opportunities.</p> <p>Demonstrate a willingness to assume managerial responsibilities.</p> <p><i>d</i> 2</p>

## 20. INFORMATION TECHNOLOGY, COMPUTER ASSISTED LEARNING AND INFORMATION MANAGEMENT

Objective: Demonstrate competence in the use and management of health information

<b>SUBJECT</b>	<b>KNOWLEDGE</b>	<b>SKILLS</b>	<b>ATTITUDES</b>
To demonstrate good use of information technology for patient care and for own personal development.	<p>Define how to retrieve and utilize data recorded in clinical systems.</p> <p>Define main local and national projects and initiatives in information technology related to genetics</p> <p>Demonstrate an understanding of the range of possible uses for clinical data and information and appreciate the dangers and benefits of aggregating clinical data.</p> <p>To understand implications of the Data Protection Act for patient confidentiality, including genetic registers.</p>	<p>Demonstrate competent use of database, word processing and statistics programmes.</p> <p>Undertake effective literature searches.</p> <p>Access genetic web sites and specialist databases to undertake searches.</p> <p>Use commercial software packages, including CYRILLIC.</p> <p>To appraise available software.</p> <p>To apply the principles of confidentiality and their implementation in terms of clinical practice in the context of information technology.</p> <p>Produce effective computer assisted presentations.</p>	<p>Demonstrate the acquisition of new attitudes in patient consultations in order to make maximum use of information technology.</p> <p>Be willing to offer advice to lay person on access to appropriate Internet sources and support groups.</p> <p>Adopt proactive and enquiring attitude to new technology.</p>
	<i>b,c,d,g,h</i> 1,2,4,8,9	<i>c,g,h,I</i> 1,2,8,9	<i>a,c,I</i> 1,4,8

## 21. THE CURRICULUM: THE TRAINING LOG

The curriculum has moved towards one that is competency-based and trainee assessment requires demonstration that skills have been acquired. The Training Log is the key document, it provides evidence of competence in Clinical Genetics. A summary of Training Log's role in the assessment process is given below.

<b>WHAT NEEDS TO BE ACHIEVED</b>	<b>LEARNING ACTIVITIES</b>	<b>ASSESSMENT</b>
A body of knowledge which every consultant clinical geneticist should know	Personal study/CPD/courses On-the-job training/use of databases Attendance at specialist clinics	Educational supervisors' reports. Written case reports Evidence of achieving required clinical experience Record of CPD
Ability to identify and solve an unfamiliar clinical problem	Use of databases and references The advice of experienced colleagues. Presentations at clinical meetings	Educational supervisors' reports Written case reports Mini CEX
Ability to communicate complex genetic information to a family and to identify important implications for the extended family	Reports from trainers and genetic co-workers. Courses on communication and counselling	Direct observation by trainer or review of videotaped consultations Mini CEX
Understanding the principles of audit	Undertaking an audit project that is written up and included in the log book	Assessed by the SAC representative at the PYA
Ability to understand the principles of research and the ability to assess the value of publications in the literature	Participation at journal clubs Undertaking a research project. Record of training in research including statistics	Trainers' reports at the PYA Publication(s) in peer-reviewed journal
Management skills	Active participation at departmental meetings Attendance on management course(s)	Reports from administrative and secretarial staff Trainers' reports
Professional behaviour/Good Medical Practice/Maintaining CPD	Reflective personal development	360° assessment