SPECIALTY TRAINING CURRICULUM

FOR

CLINICAL GENETICS

AUGUST 2010

(AMENDMENTS August 2013)

Joint Royal Colleges of Physicians Training Board

5 St Andrews Place
Regent's Park
London NW1 4LB

Telephone: (020) 79351174
Facsimile: (020)7486 4160
Email: ptb@jrcptb.org.uk
Website: www.jrcptb.org.uk

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1 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 counselling of family members. Clinical Genetics specialists generally work in multidisciplinary regional genetic centres, in close collaboration with laboratory scientists, clinical co-workers (genetic counsellors) and academic colleagues.

The CCT specialist will be able to work as a consultant specialist within the National Health Service and will have the knowledge, skills and attitudes required to do this (i.e. capable of providing a high standard of professional service).

The specialty of Clinical Genetics is constantly changing and the Clinical Geneticist must take account of new knowledge and molecular developments and alter clinical practice accordingly. S/He will be an information resource for other medical specialists. Clinical Geneticists will need a wide range of clinical skills as genetic disorders can affect people of all ages and involve all body systems. Communication skills are particularly important in explaining complex concepts and genetic test results to families enabling them to make informed decisions and choose an appropriate course of action.

The clinical geneticist works closely with clinical scientists managing cytogenetic, molecular and biochemical genetic laboratories. The clinical geneticist gives advice to other professionals such as teachers, NHS commissioners and lay organisations. Finally, clinical geneticists have an important role in public education and public debate about ethical and other diverse issues that arise from new developments in the clinical application of genetic knowledge.

Effective performance as a clinical geneticist lies in the ability to be empathetic and informative without being didactic and committed to effective advocacy on behalf of patients and families struggling to adjust to the impact of potentially life-changing, events.

2 Rationale

2.1 Purpose of the Curriculum

The purpose of this curriculum is to define the process of training and the competencies needed for the award of a certificate of completion of training (CCT) in Clinical Genetics.

The curriculum also serves to provide essential information for those considering specialty training in Clinical Genetics.

The competencies to be achieved as described within the curriculum build on core training (core medical training – CMT or acute care common stem (medicine) – ACCS (M)) which in turn build on foundation training. Clinical Genetics also has trainees entering the specialty training programme following specialty training experience in Paediatrics and Child Health. The early years of specialty training build on the competencies successfully achieved in the foundation training. This curriculum describes the competencies expected in specialty training in Clinical Genetics and how they will be attained and assessed.

The curriculum will be achieved by completing the necessary posts within educationally approved training programmes in Medicine, Child Health and Clinical Genetics. Trainees

entering ST3 in Clinical Genetics will have training experience in either Core Medical Training or in Paediatrics and Child Health or both.

The curriculum covers training for all four nations of the UK.

2.2 Development

This curriculum was developed by the Specialty Advisory Committee for Clinical Genetics under the direction of the Joint Royal Colleges of Physicians Training Board (JRCPTB). It replaces the previous version of the curriculum dated May 2007, with changes to ensure the curriculum meets GMC's standards for Curricula and Assessment, and to incorporate revisions to the content and delivery of the training programme. Major changes from the previous curriculum include the incorporation of generic, leadership and health inequalities competencies.

The SAC membership represents teachers, trainers and trainees in the specialty, and the opinions of the Clinical Genetics Society, Royal College of Paediatrics and Child Health as well as patient views are gained through their representation on the SAC. The input of each Regional Training Centre in Clinical Genetics was sought through consultation with the Regional Specialty Advisors in Clinical Genetics.

2.3 Training Pathway

Specialty training in Clinical Genetics consists of core and higher speciality training. Core training provides physicians with: the ability to investigate, treat and diagnose patients with acute and chronic medical symptoms; and with high quality review skills for managing inpatients and outpatients. Higher speciality training then builds on these core skills to develop the specific competencies required to practise independently as a consultant in Clinical Genetics

Core training may be completed in either a Core Medical Training (CMT), Acute Care Common Stem (ACCS) programme or Level 1 Paediatrics. The full curriculum for specialty training in Clinical Genetics therefore consists of the curriculum for either CMT, ACCS or Framework of Competences for Level 1 Training in Paediatrics plus this specialty training curriculum for Clinical Genetics.

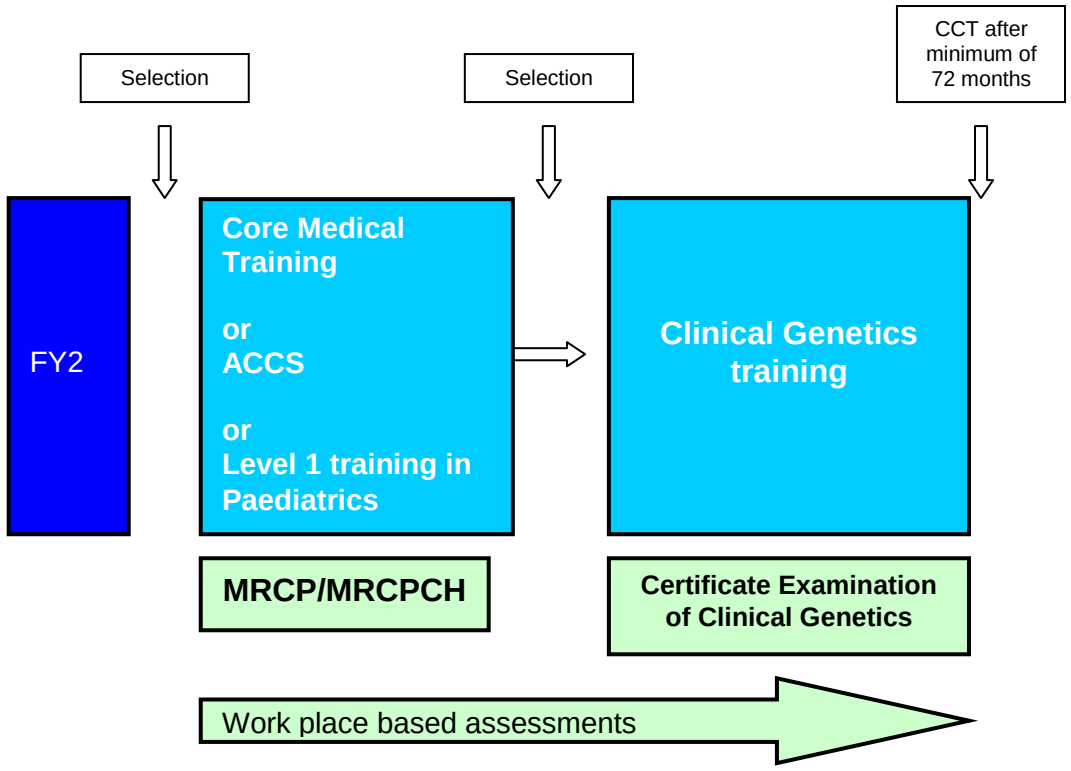
The approved curriculum for CMT is a sub-set of the Curriculum for General Internal Medicine (GIM). A "Framework for CMT" has been created for the convenience of trainees, supervisors, tutors and programme directors. The body of the Framework document has been extracted from the approved curriculum but only includes the syllabus requirements for CMT and not the further requirements for acquiring a CCT in GIM.

Core Medical training programmes are designed to deliver core training for specialty training by acquisition of knowledge and skills as assessed by the workplace based assessments and the MRCP. Programmes are usually for two years and are broad based consisting of four to six placements in medical specialties. These placements over the two years must include direct involvement in the acute medical take. Trainees are asked to document their record of workplace based assessments in an ePortfolio which will then be continued to document assessments in specialty training. Trainees completing core training will have a solid platform of common knowledge and skills from which to continue into Specialty Training at ST3, where these skills will be developed and combined with specialty knowledge and skills in order to award the trainee with a certificate of completion of training (CCT).

There are common competencies that should be acquired by all physicians during their training period starting within the undergraduate career and developed throughout the postgraduate career. These are initially defined for CMT and then developed further in the specialty. This part of the curriculum supports the spiral nature of learning that underpins a trainee's continual development. It recognises that for many of the competences outlined there is a maturation process whereby practitioners become more adept and skilled as their career and experience progresses. It is intended that doctors should recognise that the acquisition of basic competences is often followed by an increasing sophistication and complexity of that competence throughout their career. This is reflected by increasing expertise in their chosen career pathway.

For entrants to specialist training from a paediatric training route, successful completion of training including the MRCPCH examination is a requirement.

The training pathway for a Clinical Genetics trainee is shown below:



2.4 Enrolment with JRCPTB.

Trainees are required to register for specialist training with JRCPTB at the start of their training programmes. Enrolment with JRCPTB, including the complete payment of enrolment fees, is required before JRCPTB will be able to recommend trainees for a CCT. Trainees can enrol online at www.jrcptb.org.uk

2.5 Duration of Training

This curriculum is competency based thus the duration of training is determined by the time to achieve competence (assuming satisfactory progress). The SAC has advised that trainees will usually achieve competence in 6 years of a specialty training programme (2 years core training, 4 years Clinical Genetics training). The programme to which the trainee is appointed will be based in a regional genetics centre and have named educational supervisors (and consultant trainers). One consultant within the

same region will act as Programme Director. In each centre, there is a minimum of one consultant per trainee. GMC is responsible for inspection and approval of training posts within programmes. The Deanery is responsible for local quality assurance of training and ensuring that training programmes meet the GMC standards for postgraduate medical education.

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 MSc in Clinical Genetics and up to 3 months is awarded for a BSc in Genetics if approval is given in advance. 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 educational credit that is allowable for any combination of research or degree study. A full 3 years of clinically based, specialty training is the minimum.

Trainees who wish to undertake part of their clinical genetic specialty training overseas must ensure that this is in a recognised Genetics centre with clinical and educational 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. Training accreditation will be granted following completion on receipt of evidence of satisfactory progress and assessment. A minimum of two years clinical training must be undertaken in the base centre in the UK, ideally including the final 12 months of training prior to CCT. All training must be seen to be part of an approved programme; trainees who seek recognition of experience elsewhere run the risk of not completing a full CCT programme, and may have to seek specialist registration through Article 14 (Certificate of Eligibility for Specialist Registration).

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

2.6 Less than Full Time Training (LTFT)

Trainees who are unable to work full-time are entitled to opt for less than full time training programmes. EC Directive 2005/36/EC requires that:

- LTFT shall meet the same requirements as full-time training, from which it will differ only in the possibility of limiting participation in medical activities.
- The competent authorities shall ensure that the competencies achieved and the quality of part-time training are not less than those of full-time trainees.

The above provisions must be adhered to. LTFT trainees should undertake a pro rata share of the out-of-hours duties (including on-call and other out-of-hours commitments) required of their full-time colleagues in the same programme and at the equivalent stage.

EC Directive 2005/36/EC states that there is no longer a minimum time requirement on training for LTFT trainees. In the past, less than full time trainees were required to work a minimum of 50% of full time. With competence-based training, in order to retain competence, in addition to acquiring new skills, less than full time trainees would still normally be expected to work a minimum of 50% of full time. If you are returning or converting to training at less than full time please complete the LTFT application form on the JRCPTB website www.jrcptb.org.uk .

Funding for LTFT is from deaneries and these posts are not supernumerary. Ideally therefore 2 LTFT trainees should share one post to provide appropriate service cover.

Less than full time trainees should assume that their clinical training will be of a duration pro-rata with the time indicated/recommended, but this should be reviewed during annual appraisal by their TPD and chair of STC and Deanery Associate Dean for LTFT training. As long as the statutory European Minimum Training Time (if relevant), has been exceeded, then indicative training times as stated in curricula may be adjusted in line with the achievement of all stated competencies**.**

3 Content of Learning

3.1 Programme Content and Objectives

The general aim of the training programme is to enable the Clinical Geneticist to work effectively as a consultant within the NHS.

To provide a competent, caring service that is fit for purpose, underpinned by scientific understanding of genetic principles and to discharge professional duties in a timely, sensitive and patient and family-centred manner.

3.2 Good Medical Practice

In preparation for the introduction of licensing and revalidation, the General Medical Council has translated Good Medical Practice into a Framework for Appraisal and Assessment which provides a foundation for the development of the appraisal and assessment system for revalidation. The Framework can be accessed at http://www.gmc-uk.org/Framework_4_3.pdf_25396256.pdf

The Framework for Appraisal and Assessment covers the following domains:
Domain 1 – Knowledge, Skills and Performance
Domain 2 – Safety and Quality
Domain 3 – Communication, Partnership and Teamwork
Domain 4 – Maintaining Trust

The “GMP” column in the syllabus defines which of the 4 domains of the Good Medical Practice Framework for Appraisal and Assessment are addressed by each competency. Most parts of the syllabus relate to “Knowledge, Skills and Performance” but some parts will also relate to other domains.

3.3 Syllabus

In the tables below, the “Assessment Methods” shown are those that are appropriate as **possible** methods that could be used to assess each competency. It is not expected that all competencies will be assessed and that where they are assessed not every method will be used. See section 5 for more details.

Syllabus Content

1. GOOD CLINICAL CARE

History, Examination, Investigations, Safe Prescribing, Management & Note keeping Skills:

Pre-Clinic Preparation

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

Knowledge	Assessment Methods	GMP
Knowledge of relevant disorder acquired by background reading	CbD	1
Skills		
Be able to review medical records and identify information sources including databases and literature searches	CbD	1
Behaviours		
Appreciate the importance of identifying key issues and being prepared to deal with these	CbD, mini-CEX	1

History

Knowledge	Assessment Methods	GMP
Define the patterns of symptoms found in patients presenting with genetic disease	CbD, CE	1
Recognise reliable and unreliable family history data and identify sources for verification	CbD, mini-CEX	1,2
Recognises importance of different elements of history	mini-CEX	1
Recognises that patients do not present history in structured fashion	mini-CEX	1,3
Knows likely causes and risk factors for conditions relevant to mode of presentation	mini-CEX	1
Recognise that the patient's agenda and the history should inform examination, investigation and management	mini-CEX	1
Skills		
Be able to take and analyse a clinical history in a relevant, succinct and logical manner	CbD, mini-CEX	1
Be able to overcome difficulties of language, physical and mental impairment	mini-CEX, MSF, PS	1,3
Use interpreters and advocates appropriately	mini-CEX, MSF, PS	3
Elicit family history information in a sensitive and understanding manner	mini-CEX, MSF, PS	1,3,4
Draw complex pedigrees accurately, including consanguinity loops, recording appropriate information	CbD, mini-CEX	1
Manages time and draws consultation to a close appropriately	mini-CEX	1,3

Recognises that effective history taking in non-urgent cases may require several discussions with the patient and other parties, over time	mini-CEX	1,3
Supplements history with standardised instruments or questionnaires when relevant	mini-CEX	1,3
Manages alternative and conflicting views from family, carers, friends and members of the multi-professional team	mini-CEX	1,3
Assimilates history from the available information from patient and other sources including members of the multi-professional team	mini-CEX	1,3
Recognises and interprets appropriately the use of non verbal communication from patients and carers	mini-CEX	1,3
Focuses on relevant aspects of history	mini-CEX	1,3
Maintains focus despite multiple and often conflicting agendas	mini-CEX	1,3
Behaviours		
Show empathy with patients and other family members	mini-CEX, MSF, PS	3,4
Appreciate the importance of psychological and social factors of patients and relatives in genetic disease	CbD	3,4
Attention to detail and accuracy in collecting and checking family history and medical data	CbD, mini-CEX	2,3
Appreciate the confidentiality and ethical issues arising from family history gathering	CbD	4
Shows respect and behaves in accordance with Good Medical Practice	mini-CEX	3,4

Examination

Knowledge	Assessment Methods	GMP
Define the pathophysiological basis of physical signs	CbD, CE	
Define the clinical signs found in genetic diseases	CbD, CE	
Understands the need for a targeted and relevant clinical examination	CbD, mini-CEX	1
Understands the basis for clinical signs and the relevance of positive and negative physical signs	CbD, mini-CEX	1
Recognises constraints to performing physical examination and strategies that may be used to overcome them	CbD, mini-CEX	1
Recognises the limitations of physical examination and the need for adjunctive forms of assessment to confirm diagnosis	CbD, mini-CEX	1
Recognise when the offer/ use of a chaperone is appropriate or required	CbD, mini-CEX	1
Skills		
Be able to perform a reliable and appropriate examination to elicit relevant signs of genetic disease	mini-CEX	1
Perform examination appropriately in situations involving cultural sensitivity	mini-CEX, MSF, PS	1,4
Understand when additional specialist examination is required	CbD, mini-CEX	1
Performs an examination relevant to the presentation and risk factors	CbD, mini-CEX	1

that is valid, targeted and time efficient		
Recognises the possibility of deliberate harm (both self harm and harm by others) in vulnerable patients and report to appropriate agencies	CbD, mini-CEX	1,2
Behaviours		
Respect patients' dignity and confidentiality	CbD, mini-CEX, MSF, PS	4
Appropriately involve relatives	CbD	3,4
In particular ensure examination whilst clinically appropriate considers social, cultural and religious boundaries to examination, appropriately communicates and makes alternative arrangements where necessary	CbD, mini-CEX, MSF	1,4

Investigations Including Imaging

Knowledge	Assessment Methods	GMP
Know the predictive value of results of investigations	CbD, CE	1
Define the pathophysiological basis of investigations	CbD, CE	1
Define the indications for investigations	CbD, CE	1
Define the risks and benefits of investigations	CbD, CE	1
Know the cost effectiveness of individual investigation	CbD	1
Skills		
Ability to prioritise investigations and interpret the results	CbD, CE, mini-CEX	1,3
Ability to perform investigations competently where relevant	Cbd	1
Ability to liaise and discuss investigations with colleagues and to order them appropriately	CbD, MSF	1,3
Behaviours		
Willingness to explain to patient and where necessary family the rationale for investigations, and possible unwanted effects	CbD, mini-CEX, PS	3,4

Diagnosis and Management

Knowledge	Assessment Methods	GMP
Recognise pitfalls in single gene inheritance including variable expressivity and reduced penetrance, somatic and gonadal mosaicism	CbD, CE	1,2
Be able to formulate differential diagnoses for genetic disorders	CbD, CE	1
Skills		
Present genetic information to a patient in a sensitive and understanding manner	CbD, mini-CEX, MSF, PS	3,4
Calculate genetic risk in single gene disorders by hand	CbD, CE	1
Calculate genetic risk by use of a computer programme	CbD	1
Use computerized genetic databases and registers for information retrieval	CbD	1
Present undiagnosed cases to colleagues, including dysmorphology	CbD	3

club meetings		
Clearly and openly explain management options	CbD, mini-CEX	3
Behaviours		
Show appropriate attitudes towards patients and their symptoms and be conscious of religious or other philosophical contexts particularly in respect to prenatal diagnosis	CbD, mini-CEX	3,4
Sensitivity in breaking bad news	CbD, mini-CEX, MSF, PS	3,4
Appreciate the impact of diagnosing serious genetic conditions on family relationships	CbD, PS	3,4

Note- Keeping, Letters, etc

Knowledge	Assessment Methods	GMP
Define the structure, function and legal implications of medical records & medico-legal reports	CbD	1,3,4
Know the relevance of the data protection pertaining to patient confidentiality	CbD	1
Skills		
Record concisely, accurately, confidentially and legibly the appropriate elements of the history, examination, results of investigations, differential diagnosis and management plan	CbD, MSF	1,2,3,4
Behaviours		
Timely and cost effective dictation and communication with medical secretaries	MSF	2,3
Prompt and accurate communication with primary care and other agencies	MSF	2,3
Show courtesy towards other healthcare professionals	MSF	3

Time Management and Decision Making

Time Management

To demonstrate that the trainee has the knowledge, skills and attitudes to manage time and problems effectively.		
Knowledge	**Assessment Methods**	**GMP**
Understand the need to prioritise work according to urgency and importance	CbD	1
Maintains focus on individual patient needs whilst balancing multiple competing pressures	CbD	1
Understand the roles, competences and capabilities of other professionals and support workers	CbD	1
Skills		
Recognise when he/she is falling behind and re-prioritise or ask for help	MSF	2
Organise and manage workload effectively and flexibly	CbD, Mini- CEX	1,2
Make appropriate use of other professionals and support workers	CbD, mini-CEX	1,2,3
Employs techniques for improving time management	CbD	3
Behaviours		
Have realistic expectations of tasks to be completed by self and others, particularly patients and their families	MSF	1,2,3
Willingness to consult and work as part of a team	MSF	1,2,3
Identify clinical and clerical tasks requiring attention or predicted to arise	CbD, mini-CEX	1,2

Decision Making

Knowledge	**Assessment Methods**	**GMP**
Define the steps of diagnostic reasoning	CbD, mini-CEX	1,2
Interpret history and clinical signs	CbD, mini-CEX	1
Conceptualise clinical problem in a medical, psychological and familial context	CbD, mini-CEX	1
Recognise how to use expert advice, clinical guidelines and algorithms	CbD, mini-CEX	1,2
Recognise and appropriately respond to sources of information accessed by patients	CbD, mini-CEX	1
Recognise the need to determine the most effective or “least worst” treatment both for the individual patient and for a patient cohort	CbD, mini-CEX	1,2
Define the concepts of disease natural history and assessment of risk	CbD, mini-CEX	1
Describe commonly used statistical methodology	CbD, mini-CEX	1
Know how relative and absolute risks are derived and the meaning of the terms predictive value, sensitivity and specificity in relation to diagnostic tests	CbD, mini-CEX	1

Skills		
Interpret clinical features, their reliability and relevance to clinical scenarios including recognition of the breadth of presentation of common disorders	CbD, mini-CEX	1
Incorporates an understanding of the psychological and social elements of clinical scenarios into decision making	CbD, mini-CEX	1
Construct a concise and applicable problem list using available information	CbD, mini-CEX	1
Construct an appropriate management plan in conjunction with the patient, carers and other members of the clinical team and communicate this effectively to the patient, parents and carers securing their agreement to the course of action	CbD, mini-CEX	1,3,4
Define the relevance of an estimated risk of a future event to an individual patient	CbD, mini-CEX	1
Use risk calculators appropriately	CbD, mini-CEX	1
Apply quantitative data of risks and benefits of screening and therapeutic intervention to an individual patient	CbD, mini-CEX	1,3
Search and comprehend medical literature to guide reasoning	AA, CbD	1
Generate hypothesis within context of clinical likelihood	CbD, mini-CEX	1
Test, refine and verify hypotheses	CbD, mini-CEX	1
Develop problem list and action plan	CbD, mini-CEX	1
Behaviours		
Show willingness to discuss intelligibly with a patient the notion and difficulties of prediction of future events, and benefit/risk balance of therapeutic intervention	CbD, mini-CEX	3
Show willingness to adapt and adjust approaches according to the beliefs and preferences of the patient and/or carers	CbD, mini-CEX	3
Show willingness to search for evidence to support clinical decision making	CbD, mini-CEX	1,4
Demonstrate ability to identify one's own biases and inconsistencies in clinical reasoning	CbD, mini-CEX	1,3

2. PROCEDURES

Phlebotomy

To demonstrate proficiency in clinical procedures related to genetics.		
Knowledge	**Assessment Methods**	**GMP**
Knowledge of technique	mini-CEX	1
Skills		
Ability to take blood samples from adults and children, including those with special needs	mini-CEX	1,3
Behaviours		
Understand the stress of the technique and obtain consent	mini-CEX	1,3

Skin Biopsy

Knowledge	**Assessment Methods**	**GMP**
Knowledge of technique and indications for use	mini-CEX	1
Skills		
Demonstrate ability to obtain samples suitable for analysis	mini-CEX	1,3
Behaviours		
Explain procedure appropriately and obtain consent	mini-CEX	1,3

Clinical Photography

Knowledge	**Assessment Methods**	**GMP**
Knowledge of technique	mini-CEX	1
Understand importance and confidentiality of photographic records	mini-CEX	1,3
Skills		
Demonstrate ability to take photographs of sufficient quality for clinical use	mini-CEX	
Use of digital photography and storage of data	mini-CEX	1,3
Behaviours		
Explain the need for clinical photography and obtain consent	mini-CEX	1,3

3. COMMUNICATION SKILLS AND GENETIC COUNSELLING

Within a Consultation

Acquire and demonstrate effective communication with patients, relatives and colleagues along with the habit of reflection on personal genetic counselling style and effectiveness. ("counselling" in this context means the transmission of information about genetic disease, risk and reproductive options).		
	Assessment	**GMP**

Knowledge	Methods	
How to structure a consultation appropriately	CbD, mini-CEX, PS	1,3
The importance of the patient's background, culture, education and preconceptions (beliefs, ideas, concerns, expectations) to the process	CbD, mini-CEX, PS	1,3
Be aware of social and cultural issues and practices such as:		
• The impact of cultural beliefs and practices on health outcomes	CbD, mini-CEX	1,3,4
• Health determinants that affect patients and communities	MSF, PS	
• effects of social and cultural issues on access to healthcare, including an understanding of health	MSF, PS	
• issues of migrants and refugees	MSF, PS	
Specific techniques and methods that facilitate effective and empathic communication	CbD, mini-CEX, MSF	1,3,4
Understand the importance of the developmental stage when communicating with adolescents and young adults	CbD, mini-CEX, PS	1
Skills		
Be able to communicate effectively, both verbally and in writing to patients whose first language may not be English in a manner that they understand	CbD, mini-CEX, MSF	3
Give clear information and feedback to patients and share information with relatives when appropriate	CbD, mini-CEX, PS	3
Establish a rapport with the patient and relevant others	CbD, mini-CEX, PS	1,3
Listen actively and question sensitively to guide the patient and to clarify information in particular with regard to matters that they may find it difficult to discuss, e.g. domestic violence or other abuse	mini-CEX, PS	1, 3
Utilise open and closed questioning appropriately	CbD, mini-CEX, PS	1,3
Listen actively and question sensitively to guide the patient and to clarify information	mini-CEX, PS	1,3
Identify and manage communication barriers, tailoring language to the individual patient and others and using interpreters when indicated	CbD, mini-CEX, PS	1, 3
Deliver information compassionately, being alert to and managing their and your emotional response (anxiety, antipathy etc.)	CbD, mini-CEX, PS	1,3,4
Use, and refer patients to, appropriate written and other evidence based information sources	CbD, mini-CEX, PS	1,3
Check the patient's/carer's understanding, ensuring that all their concerns/questions have been covered	CbD, mini-CEX, PS	1,3
Indicate when the consultation nearing its end and conclude with a summary and appropriate action plan; ask the patient to summarise back to check his/her understanding	CbD, mini-CEX, PS	1,3
Make accurate contemporaneous records of the discussion	CbD, mini-CEX, PS	1,3
Manage follow-up effectively and safely utilising a variety of methods (e.g. phone call, email, letter)	CbD, mini-CEX, PS	1
Ensure appropriate referral and communications with other healthcare professional resulting from the consultation are made accurately and in a timely manner	CbD, mini-CEX, PS	1

Respect diversity and recognise the benefits it may bring, as well as associated stigma	CbD, mini-CEX, PS	1,3,4
Behaviours		
Approach the situation with courtesy, empathy, compassion and professionalism, especially by appropriate body language and endeavouring to ensure an appropriate physical environment	CbD, mini-CEX, MSF, PS	1,3,4
Ensure that the approach is inclusive and patient centred and respect the diversity of values in patients, carers and colleagues	CbD, mini-CEX, MSF, PS	1,3
Be willing to provide patients with a second opinion	CbD, mini-CEX, MSF, PS	1,3
Accept uncertainty and use different methods of ethical reasoning to come to a balanced decision where complex and conflicting issues are involved	CbD, mini-CEX, MSF	1,3
Demonstrate: • Recognising good advice and continuously promoting values based non prejudicial practice	MSF	3
• Using authority appropriately and assertively; willing to follow when necessary	MSF	3

Breaking Bad News

Knowledge	Assessment Methods	GMP
Know how to structure the interview and where it should take place	CbD, mini-CEX	
Be aware of the normal bereavement process and behaviour	CbD	
How bad news is delivered irretrievably affects the subsequent relationship with the patient	CbD, mini-CEX, MSF, PS	1
Every patient may desire different levels of explanation and have different responses to bad news	CbD, mini-CEX, PS	1,4
That bad news is confidential but the patient may wish to be accompanied	CbD, mini-CEX, PS	1
Breaking bad news can be extremely stressful for the doctor or professional involved	CbD, mini-CEX	1,3
“Bad news” may be expected or unexpected and it cannot always be predicted	CbD, mini-CEX	1,3
Sensitive communication of bad news is an essential part of professional practice	CbD, mini-CEX	1,3
“Bad news” has different connotations depending on the context, individual, social and cultural circumstances	CbD, mini-CEX, PS	1,3
Skills		
Be able to break bad news in steps appropriate to the understanding of the individual and be able to support distress	MSF	1,3
Demonstrate to others good practice in breaking bad news	CbD, MSF	1,3
Recognises the impact of the bad news on the patient, carer, supporters, staff members and self	CbD, MSF	1,3,4
Encourage questioning and ensure comprehension	CbD, MSF	1,3
Respond to verbal and visual cues from patients and relatives	CbD, MSF	1,3

Act with empathy, honesty and sensitivity avoiding undue optimism or pessimism	CbD, MSF	1,3
Structures the interview inappropriately	CbD, MSF	1,3
Behaviours		
Show empathy, honesty and sensitivity	MSF	4
Show leadership in breaking bad news	CbD, DOPS, MSF	1
Respect the different ways people react to bad news	CbD, DOPS, MSF	1
Ensure appropriate recognition and management of the impact of breaking bad news on the doctor	MSF	2

Specific Genetic Issues

Knowledge	Assessment Methods	GMP
Knowledge of ethnic difference in the incidence of genetic disease	CE	1
Understanding of cross-cultural issues including consanguinity and arranged marriages	CbD, CE, mini-CEX	1
Understanding of religious beliefs and attitudes to prenatal diagnosis and assisted reproduction techniques	CbD, mini-CEX	1
Skills		
Use of “non-directive” counselling skills	CbD, mini-CEX, MSF, PS	1,3
Effective use of co-counsellors	CbD, mini-CEX, MSF, PS	1,3
Communication of genetic information and risk to children and adolescents	CbD, mini-CEX, MSF	1,3
Communication with adults and children with learning disability	CbD, mini-CEX, MSF	1,3,4
Recognising which patients will benefit from referral on to psychological services	CbD	1
Behaviours		
Appreciate patient and family anxieties, both rational and irrational	CbD, mini-CEX	1,3,4
Appreciate that every person is influenced by their own culture, ethnicity and beliefs	CbD, CE, mini-CEX	1,3
Appreciate the importance of genetic counsellors	CbD, MSF	1,3,4
Cultivate habit of reflection and discussion with colleagues after counselling sessions	CbD, MSF	1,3,4
Readiness to alter practice in light of experience and feed-back	CbD	2,4

Complaints

Knowledge	Assessment Methods	GMP
Be aware of the local complaints procedures	MSF	1,2
Be aware of systems of independent review	MSF	1,2
Recognise factors likely to lead to complaints (poor communication, dishonesty, clinical errors, adverse clinical outcomes etc.)	CbD, MSF	1,2

Recognise the impact of complaints and medical error on staff, patients, and the National Health Service	CbD, DOPS, MSF	1,3
Skills		
Manage dissatisfied patients / relatives	MSF	1,2
Contribute to processes whereby complaints are reviewed and learned from	CbD, DOPS, MSF	1,2,3
Explain comprehensibly to the patient the events leading up to a medical error or serious untoward incident, and sources of support for patients and their relatives	CbD, DOPS, MSF	1,2,3,4
Deliver an appropriate apology and explanation (either of error of for process of investigation of potential error and reporting of the same)	CbD, DOPS, MSF	1,2,3,4
Distinguish between system and individual errors (personal and organisational)	CbD, DOPS, MSF	1,2
Show an ability to learn from previous error	CbD, DOPS, MSF	1,2,4
Recognise when something has gone wrong and identify appropriate staff to communicate this with	CbD	2
Behaviours		
Act with honesty and sensitivity and promptly	CbD, MSF	
Be prepared to accept responsibility	CbD, MSF	
Take leadership over complaint issues	CbD, DOPS, MSF	1
Recognise the impact of complaints and medical error on staff, patients, and the National Health Service	CbD, DOPS, MSF	1,3
Contribute to a fair and transparent culture around complaints and errors	CbD, DOPS, MSF	1,4
Adopt behaviour likely to prevent causes for complaints	CbD	2

4. FORMAL GENETICS AND BASIC SCIENCES

Understand cellular and molecular mechanisms that underpin inheritance in man
Identify the social and ethical implications of genetic knowledge
Understand patterns of inheritance and undertake risk assessment
Have knowledge of emerging genetic technologies and their application (including gene therapy)

Knowledge	Assessment Methods	GMP
The chromosomal basis of heredity (mitosis and meiosis)	CE	1
Mechanisms of origin of numerical and structural chromosome abnormalities	CE	1
Behaviour of structural chromosome abnormalities at meiosis	CE	1
The chemical structure of DNA and replication	CE	1
Central dogma of cell biology: transcription and translation.	CE	1
Modes of inheritance (Mendelian and non Mendelian)	CbD, CE	1
Risk calculations including combinatorial probability and Bayes Theorem	CbD, CE	1
The clinical embryology and molecular mechanisms of human malformation syndromes	CbD, CE	1
Principles of teratogenesis and pregnancy associated risks	CbD, CE	1
Mechanisms of mutagenesis and estimation of mutation rates	CbD, CE	1
History of genetics	CE	1
Skills		
Recognition of different inheritance patterns in pedigrees.	CbD, CE	1
Pedigree-based calculation of segregation ratios for structural chromosome abnormalities	CbD, CE	1
Empiric risk calculations (occurrence and recurrence risks).	CbD, CE	1
Perform Bayesian risk calculations including linkage-based risk calculations	CE	1
Calculate gene frequencies – understand the implications of the Hardy-Weinberg equilibrium	CE	1
Apply knowledge to interpret results of chromosome and molecular genetic analysis	CbD, CE	1
Behaviours		
Commitment to lifelong self-directed learning	MSF	2,4
Appreciation the impact of genetic disorders on individuals and families	CbD	2,4
Appreciate potential benefits and harm of new genetic technologies	CbD	2,4
Appreciate public concerns about the application of new genetic technologies	CbD	2,4

5. COMMON GENETIC REFERRALS

To provide the trainee with the skills and knowledge to be able to carry out specialist diagnosis, assessment and genetic counselling genetic conditions		
Knowledge	**Assessment Methods**	**GMP**
The genetic basis and clinical features of common genetic conditions	CbD, CE	1
The medical and surgical complications of common genetic conditions and indications for referral for specialist opinion	CbD, CE	1
Molecular/cytogenetic testing that is available and its application to diagnosis, predictive testing, carrier testing and prenatal diagnosis	CbD, CE	1
Application and limitations of current tests	CbD, CE	1
Knowledge of current clinical treatments for 'core' conditions and gene therapy trial	CbD, CE	1
Skills		
Be able to take a relevant history, perform an appropriate examination and formulate clinical diagnoses	CbD, mini-CEX	1
Be able to assess patients and families affected by genetic conditions	CbD	1
Judge when it is necessary to sustain supportive relationships with patients with chronic disease	CbD	1,4
Be able to discuss reproductive options (AID, ICSI, IVF, pre-implantation diagnosis) with the patient and their partner in a sensitive manner	CbD, mini-CEX	1,3
Be able to discuss and formulate integrated care pathways and management plans with individuals/families	CbD, mini-CEX	1,3
Verify diagnoses from old hospital records	CbD	1
Behaviours		
Value the contribution and role of other specialists	CbD	1
Appreciate role of patient education and support groups e.g. in type 1 neurofibromatosis	CbD	1
Appreciate the role of the general practitioner in management of chronic disease	CbD	1
Apply good clinical care and counselling skills	CbD, mini-CEX	1

6. NEUROGENETICS

To provide the trainee with the skills and knowledge to recognise genetic causes of central and peripheral nervous system dysfunction		
Knowledge	**Assessment Methods**	**GMP**
Classification and molecular basis of common genetic neuromuscular disorders	CbD, CE	1
Genetic aspects and clinical presentation of trinucleotide repeat disorders	CbD, CE	1
Basic neuropathology and differential diagnosis of hereditary dementias	CbD, CE	1
Mitochondrial diseases – clinical, biochemical and genetic features	CbD, CE	1
Genetic causes of mental retardation (static and progressive)	CbD, CE	1
Genetic contribution to autism and autistic spectrum disorders	CbD, CE	1
Genetic contribution to psychiatric disease in adults	CbD, CE	1
Skills		
Recognise family history data that suggest familial neurological disease	CbD, mini-CEX	1
Be able to confirm clinical signs in affected individuals	CbD, mini-CEX	1
Be able to draw up a differential diagnosis and institute appropriate genetic testing	CbD, CE, mini-CEX	1
Assessment of symptoms and signs in patients at risk of adult-onset neurogenetic disease	CbD, mini-CEX	1
Application of protocols for pre-symptomatic diagnosis of Huntington's disease and other neurodegenerative disorders	CbD, mini-CEX	1
Make timely, appropriate referrals to other specialists such as neurologists, psychologists, psychiatrists, speech therapists	CbD, MSF	1
Behaviours		
Appreciation of family stress caused by risk or eventuality of neurodegeneration	CbD, MSF	1,3,4
Appreciate social problems encountered by adults with mild/moderate learning disability	CbD, MSF	1,3
Appreciate issues involved in predictive testing	CbD, MSF	1,3

7. PAEDIATRIC GENETICS AND DYSMORPHOLOGY

To provide the trainee with the skills and knowledge to make syndrome diagnosis in children		
Knowledge	**Assessment Methods**	**GMP**
Identify normal developmental milestones and diagnose delayed development	CbD, CE, mini-CEX	1
Explain morphogenesis in terms of deformation, malformation, disruption and dysplasia	CbD, CE	1
Have knowledge of common and rarer dysmorphic syndromes	CbD, CE	1
Skills		
Be able to take a relevant history, and perform an appropriate examination, obtain illustrative photographs	CbD, mini-CEX	1
Have a rational approach to investigation of children with delayed development and/or dysmorphic syndromes.	CbD, mini-CEX	1
Formulate differential diagnoses of unknown syndromes	CbD, CE, mini-CEX	1
Utilise journals and databases used in syndrome identification	CbD	1
Cultivate critical assessment of database information and case reports to identify uncertainty and subjectivity in syndrome diagnosis	CbD	1
Be able to provide a diagnostic service within a multidisciplinary clinical team	CbD, mini-CEX, MSF	1
Present and discuss cases with colleagues	CbD	1
Behaviours		
Recognise importance of clinical judgement, timing, and tact when diagnosing and informing parents of an infant with serious malformation or handicap	CbD, MSF, PS	1,3,4
Appreciate the emotional reactions of parents following early diagnosis of syndrome or recognition of developmental delay	CbD, MSF, PS	1,3,4
Appreciate the adverse reaction families may experience following retraction of a previous diagnosis	CbD, MSF, PS	1,3,4
Recognise and explain to families when diagnostic work crosses the boundary into research and the constraints that this imposes	CbD	1,3,4

8. CANCER GENETICS

Ability to diagnose rare cancer syndromes and recognise when common cancers are likely to have a single gene basis

Ability to recommend targeted screening in individuals who are identified as having increased risk

Coordination of appropriate molecular genetic testing

Knowledge	Assessment Methods	GMP
The genetic and environmental factors that affect risk of developing cancer	CE	1
Current recommendations concerning tumour surveillance in cancer-prone families	CbD, CE	1
Knowledge of clinical features of genetic cancer syndromes	CbD, CE	1
Genetic mechanisms in neoplasia: Knudson's two-hit hypothesis	CE	1
Knowledge of molecular basis of cancer genetic syndromes	CE	1
Knowledge of how inherited and environmental predisposition may affect cancer treatment	CbD, CE	1
Skills		
Be able to take a relevant history, perform an appropriate examination and undertake risk estimation using a variety of methods	CbD, mini-CEX	1
Use of cancer registers and other sources to verify diagnoses	CbD	1
Use disease registers to support follow-up of affected and at-risk patients	CbD	1
Assessment of screening protocols for at-risk relatives	CbD	1
Identify at-risk patients and relatives who are eligible to participate in trials of cancer prevention strategies	CbD, CE, mini-CEX	1
Behaviours		
Demonstrate awareness of the roles primary care and genetic associates play in assessing families where relatives are at risk of developing cancer	CbD, MSF	1,3
Inform patients about lifestyle factors that affect cancer risk	CbD, mini-CEX, PS	1,3
Support general practitioners with the long-term management of selected patients with familial cancer syndromes	CbD	1,3
Liaise with other specialists as appropriate e.g. for advice about prophylactic mastectomy and work as a member of a multidisciplinary team	CbD, MSF	1,2,3
Understand the impact of cancer risk on individuals and families	CbD, MSF, PS	1,2,3

9. PRENATAL DIAGNOSIS AND FETAL DYSMORPHOLOGY

To provide the trainee with the skills and knowledge to undertake genetic assessment of actual and potential problems in the foetus, and provide parents with advice about prognosis and inheritance

To acquire the skills and knowledge to assess the risk of potential genetic problems in the foetus prior to pregnancy and to advise parents about the options and procedures open to them within the current legal framework

To develop the skills and knowledge to assess foetal abnormality during pregnancy, to provide parents with information about prognosis, genetic investigations, including post-mortem examination and storage of foetal tissue

Knowledge	Assessment Methods	GMP
Understand the natural history of prenatally diagnosed conditions, including common single gene and chromosome abnormalities	CbD, CE, mini-CEX	1,2
Know the indications for and methods of preimplantation and prenatal diagnosis	CbD, CE, mini-CEX	1,2
Be informed of the latest advances in prenatal diagnosis such as testing free foetal DNA in maternal blood and the potential for non-invasive prenatal DNA diagnosis	CbD, CE, mini-CEX	1,2
Knowledge of the law pertaining to termination of pregnancy for foetal abnormality	CbD, CE, mini-CEX	1,2
Know the indications for, process and limitations of foetal post-mortem examination and issues of consent	CbD, CE, mini-CEX	1,2
Have knowledge of RCPath guidelines on retention and storage of foetal tissues and the Human Tissues Act	CbD, CE, mini-CEX	1,2
Skills		
Interpret family history data	CbD, mini-CEX	1,3
Provide genetic advice for women who may undergo preimplantation or prenatal diagnosis	CbD, mini-CEX	2,3
Formulate differential diagnoses and assess prognosis in collaboration with the foetal medicine team	CbD, mini-CEX	2,3
Assess risk to foetus when pregnancies are exposed to hazards such as congenital infections, alcohol, ionising irradiation or drugs	CbD, mini-CEX	2,3
Assess clinical significance of chromosome, DNA and foetal imaging in the context of foetal abnormality	CbD, mini-CEX	2,3
Evaluate foetal post-mortem findings	CbD, mini-CEX	2,3
Behaviours		
Appreciate the advantages and disadvantages of preimplantation and prenatal diagnosis in each situation	CbD	1,3
Non-judgmental appreciation of the ethical and religious dimensions to preimplantation and prenatal diagnosis	CbD	1,3,4
Awareness of the adverse psychological effects of termination of pregnancy for fetal abnormality	CbD	1,3
Appreciate the role of relevant patient support groups and other counselling services	CbD	1,3,4

10. CARDIAC GENETICS

Ability to diagnose inherited cardiac conditions (ICC)

Ability to recommend targeted screening in individuals who are identified as having increased risk of an ICC

Ability to coordinate appropriate molecular genetic testing

Knowledge	Assessment Methods	GMP
Knowledge of clinical features of ICC syndromes, including Marfan syndrome and related disorders	CbD, CE, mini-CEX	1
Knowledge of molecular basis of ICC syndromes	CbD, CE	1
Current recommendations concerning cardiac surveillance in ICC families	CE	1
Knowledge of genetic causes of sudden adult death	CbD, CE	1
Skills		
Be able to take a relevant history, perform an appropriate examination	mini-CEX	1
Work with bereaved families following sudden adult death	MSF	1,3,4
Use of Ghent criteria for diagnosing Marfan syndrome	CE, mini-CEX	1
Assessment of screening protocols for at-risk relatives	CbD, mini-CEX	1,2
Coordinate diagnostic and predictive genetic testing in ICC families	mini-CEX	1,2
Identify at-risk patients and relatives who are eligible to participate in prevention strategies (e.g. therapeutic trials)	CbD	1,2
Behaviours		
Demonstrate awareness of the roles of primary care, specialist nurses and genetic counsellors play in assessing families where relatives are at risk of developing ICC	MSF	3
Inform patients about lifestyle factors that affect risk	CbD, mini-CEX	3
Support primary and secondary care professionals with the long-term management of selected patients with ICC syndromes	CbD, mini-CEX	1,3
Work as a member of a multidisciplinary team	MSF	3,4
Understand the impact of ICC risk on individuals and families; and demonstrate awareness of psychological impact of sudden adult death	CbD	2,3

11. LABORATORY GENETICS

To acquire skills and knowledge to interpret genetic laboratory results within a clinical setting, by completing an attachment in the genetic laboratories		
Knowledge	**Assessment Methods**	**GMP**
Techniques for conventional chromosome analysis in different tissues	CE	1
Laboratory techniques and application of new cytogenetic tests e.g. FISH/CGH	CE	1
Use of ISCN nomenclature	CE	1
Molecular genetic techniques in common usage – (DNA extraction, Southern Blotting, PCR, DNA sequencing)	CE	1
Application of DNA-based testing, for gene mapping, linkage and mutation detection	CE	1
Potential application of new DNA technologies	CE	1
Sensitivity and specificity of laboratory tests	CE	1
Investigative approach to biochemical diagnosis of inborn errors of metabolism (via experience gained at metabolic disease clinics)	CbD, CE	1
The operation of local and national antenatal and newborn, genetic disease screening programmes	CbD, CE	1
Skills		
Interpretation of clinical consequences of abnormal karyotypes, enzyme deficiencies and molecular test results	CbD, CE	1,2,3
Liaise with molecular and cytogenetics scientists in analysis of test results	MSF	1,3
Provide advice to laboratory on the wording of reports to referring clinicians	CbD, MSF	1,2,3,4
Genetic risk calculation based on laboratory test results (e.g. MLINK, Bayesian analysis)	CE	1,2,3
Behaviours		
Awareness of the importance of informed consent that arise in relation to storage of DNA samples and cell lines	CbD, mini-CEX	1,3
Willingness to liaise with colleagues to interpret laboratory results.	MSF	3
Recognise the value of clear reports and the importance of communicating results clearly to families	CbD	1,3

12. MAINTAINING TRUST

Professional Behaviour

Continuity of Care

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

Knowledge	Assessment Methods	GMP
Understand the relevance of continuity of care	CbD	
Know main methods of ethical reasoning	CbD	1
Define the concept of modern medical professionalism	CbD	1
Outline the relevance of professional bodies (Royal Colleges, JRCPTB, GMC, Postgraduate Dean, BMA, specialist societies, medical defence societies)	CbD	1
Skills		
Make adequate arrangements to cover leave.	MSF	2,3
Practise with professionalism including integrity, compassion, altruism, continuous improvement, aspiration to excellence, respect of cultural and ethnic diversity, and with regard to the principles of equity	CbD, mini-CEX, MSF, PS	1,2,3,4
Work in partnership with patients and members of the wider healthcare team	CbD, mini-CEX, MSF	3,4
Liaise with colleagues to plan and implement work rotas	MSF	3
Promote awareness of the doctor's role in utilising healthcare resources optimally and within defined resource constraints	CbD, mini-CEX, MSF	1,3
Recognise and respond appropriately to unprofessional behaviour	CbD	1
Be able to handle enquiries from the press and other media effectively	CbD, DOPS	1,3
Eliminate discrimination against patients from diverse backgrounds including age, gender, race, culture, disability and sexuality	CbD	1,3
Behaviours		
Recognise the importance of punctuality and attention to detail	CbD, MSF	2,3
Recognise personal beliefs and biases and understand their impact on the delivery of health services	CbD, mini-CEX, MSF	1,4
Show willingness to act as a leader, mentor, educator and role model	CbD, mini-CEX, MSF	1,3
Be willing to accept mentoring as a positive contribution to promote personal professional development	CbD, mini-CEX	1,3
Participate in professional regulation and professional development	CbD, mini-CEX, MSF	1,2,3
Respect the rights of children, elderly, people with physical, mental, learning or communication difficulties	CbD	1,3
Behave with honesty, probity and sensitivity in a non-confrontational manner	MSF	2,3

Doctor-Patient Relationship

Knowledge	Assessment Methods	GMP
Understand all aspects of a professional relationship.	MSF	1,3
Establish the limiting boundaries surrounding the consultation.	CbD, mini-CEX	1,3
Outline health needs of particular populations e.g. ethnic minorities and recognise the impact of health beliefs, culture and ethnicity in presentations of physical and psychological conditions	CbD	1,3,4
Demonstrate how Individual behaviours impact on others; personality types, group dynamics, learning styles, leadership styles	Cbd, MSF	3,4
Skills		
Develop a relationship that facilitates solutions to patient's problems	CbD, mini-CEX, MSF	1,3,4
Deal appropriately with behaviour falling outside the boundary of the agreed doctor patient relationship in patients, e.g. aggression, violence, sexual harassment	CbD, mini-CEX, MSF	1,3,4
Develop a self-management plan with the patient	CbD, mini-CEX	1,3,4
Support patients, parents and carers where relevant to comply with management plans	CbD, mini-CEX, PS	3,4
Encourage patients to voice their preferences and personal choices about their care	mini-CEX, PS	3,4
Use assessment, appraisal, complaints and other feedback to discuss and develop an understanding of own development needs.	MSF, PS	2,4
Behaviours		
Recognise the duty of the medical professional to act as patient advocate	CbD, mini-CEX, MSF, PS	3, 4
Demonstrate:	CbD	3,4
• Acceptance of professional regulation remove bullets and merge		
• Promotion of professional attitudes and values	CbD	3,4
• Probity and the willingness to be truthful and to admit errors	CbD	3,4

Recognising Own Limitations

Knowledge	Assessment Methods	GMP
Know the extent of one's own limitations and the limitations of self professional competence and know when to ask for advice.	CbD	2,3,4
Recognise that personal beliefs and biases exist and understand their impact (positive and negative) on the delivery of health services	CbD	2,3,4
Skills		
Reflection on individual practice.	CbD	2,3,4
Behaviours		
Be willing to consult and to admit mistakes.	CbD, MSF	2,3,4
Be confident and positive in one's own professional values	CbD, MSF	2,3,4
Be aware of one's own behaviour and how it might impact on	CbD, MSF	2,3,4

patients' health issues

Stress & Personal Health

Knowledge	Assessment Methods	GMP
Know the effects of stress and tools and techniques for managing it	MSF	2,4
Demonstrate knowledge of the role and responsibility of occupational health and other support networks for doctors	MSF	2,4
Know about one's responsibilities to the public	CbD	1
Skills		
Develop appropriate coping mechanisms for stress and ability to seek help if appropriate	MSF	2,4
Demonstrate the ability to recognise the manifestations of stress on self and others and know where and when to look for support	MSF	2,4
DeoBalance personal and professional roles and responsibilities. Prioritise tasks, having realistic expectations of what can be completed by self and others	MSF	2,4
Behaviours		
Being conscientious, able to manage time and delegate	MSF	2,4
Recognise personal health as an important issue	MSF	2,4
Recognise personal health as an important issue.	MSF	1

Life-Long Learning

To inculcate the habit of life-long learning		
Knowledge	**Assessment Methods**	**GMP**
Define continuing professional development	CbD	1
Skills		
Recognise and use learning opportunities.	CbD	1
Use the potential of study leave to keep oneself up to date	CbD	1
Behaviours		
Be: • self motivated • eager to learn	CbD, MSF	
Show: • willingness to learn from colleagues • willingness to accept criticism	CbD, MSF	

13. ETHICS AND LEGAL ISSUES

Principles of Medical Ethics and Confidentiality

To know, understand and apply appropriately the principles, guidance and laws regarding medical ethics and confidentiality

Knowledge	Assessment Methods	GMP
Demonstrate knowledge of the principles of medical ethics	CbD, mini-CEX	1
Outline and follow the guidance given by the GMC on confidentiality	CbD, mini-CEX	1
Define the provisions of the Data Protection Act and Freedom of Information Act	CbD, mini-CEX	1
Define the principles of Information Governance	CbD, mini-CEX	1
Define the role of the Caldicott Guardian and Information Governance lead within an institution, and outline the process of attaining Caldicott approval for audit or research	CbD, mini-CEX	1,4
Outline situations where patient consent, while desirable, is not required for disclosure e.g. serious communicable diseases, public interest	CbD, mini-CEX	1,4
Outline the procedures for seeking a patient's consent for disclosure of identifiable information	CbD, mini-CEX	1
Recall the obligations for confidentiality following a patient's death	CbD, mini-CEX	1,4
Recognise the problems posed by disclosure in the public interest, without patient's consent	CbD, mini-CEX	1,4
Recognise the factors influencing ethical decision making: including religion, personal and moral beliefs, cultural practices	CbD, mini-CEX	1
Outline the principles of the Mental Capacity Act	CbD, mini-CEX	1
Demonstrate an understanding of adolescents' and young adults' right to confidentiality and the importance of safeguarding	ACAT, CbD, mini-CEX	1
Skills		
Use and share information with the highest regard for confidentiality, and encourage such behaviour in other members of the team, whilst recognising that the familial nature of genetics means that respecting individual confidentiality can be more complex.	CbD, mini-CEX, MSF	1,2,3
Use and promote strategies to ensure confidentiality is maintained e.g. anonymisation	CbD	1
Counsel patients on the need for information distribution within members of the immediate healthcare team	CbD, MSF	1,3
Behaviours		
Encourage informed ethical reflection in others	CbD, MSF	1
Show willingness to seek advice of peers, legal bodies, and the GMC in the event of ethical dilemmas over disclosure and confidentiality	CbD, mini-CEX, MSF	1
Respect patient's requests for information not to be shared, unless this puts the patient, or others, at risk of harm	CbD, mini-CEX, PS	1,4
Show willingness to share information about their care with patients,	CbD, mini-CEX	1,3

unless they have expressed a wish not to receive such information

Informed Consent

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.

Knowledge	Assessment Methods	GMP
Know the process for gaining informed consent	CbD, CE, mini-CEX	1,3
Understand process of consent for tissue/sample storage and use	CbD, CE	1,3
How to gain consent for a research project	CbD, mini-CEX	1,3
Outline the guidance given by the GMC on consent	CbD, DOPS, MSF	1
Skills		
Present all information to patients (and carers) in a format they understand, checking understanding and allowing time for reflection on the decision to give consent with appropriate use of written material	CbD, mini-CEX, PS	1, 3
Provide a balanced view of all care options	CbD, mini-CEX, PS	1,3,4
Behaviours		
Respect a patient's rights of autonomy even in situations where their decision might put them at risk of harm	CbD, mini-CEX, PS	1
Do not withhold information relevant to proposed care or treatment in a competent patient	CbD, mini-CEX	1,3,4
Does not seek to obtain consent for procedures in which they are not competent to perform, in accordance with GMC/regulatory	CbD, mini-CEX	1,3
Show willingness to seek advance directives		
Show willingness to obtain a second opinion, senior opinion, and legal advice in difficult situations of consent or capacity	CbD, mini-CEX, MSF	1,3
Inform a patient and seek alternative care where personal, moral or religious belief prevents a usual professional action	CbD, mini-CEX, PS	1,3,4

Legal Framework for Practice

To 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

Knowledge Knowledge	Assessment Methods	GMP
All decisions and actions must be in the best interests of the patient	CbD, mini-CEX	1
Understand sources of medical legal information	CbD, mini-CEX	1
Understand disciplinary processes in relation to medical malpractice	CbD, mini-CEX, MSF	1
Skills		
Ability to cooperate with other agencies with regard to legal requirements	CbD, mini-CEX	1
Practice and promote accurate documentation within clinical practice	CbD, mini-CEX	1,3

Behaviours		
Show willingness to seek advice from the employer, appropriate legal bodies (including defence societies), and the GMC on medico-legal matters	CbD, mini-CEX, MSF	1
Promote informed reflection on legal issues by members of the team	CbD, mini-CEX, MSF	1,3
All decisions and actions must be in the best interests of the patient		

14. ORGANISATION AND PROVISION OF GENETICS SERVICES FOR POPULATIONS

To identify practical, legal and ethical issues arising from operation of genetic registers
To know the criteria against which screening programmes for genetic diseases and susceptibilities are judged

Knowledge	Assessment Methods	GMP
The genetic characteristics of populations, common gene frequencies and disease prevalence	CE	1,2
The factors that influence decisions to instigate programmes of population screening for genetic diseases	CE	1,2
Define sensitivity, specificity, and predictive values of screening tests.	CE	1,2
Knowledge of current screening programmes	CE	1,2
Knowledge of appropriate population-based registers	CE	1,2
Skills		
Team-working with database managers, genetic associates and nurse specialists in:	MSF	2,3
• '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)	MSF, CE	2,3
• family based screening for individuals at high risk of developing cancer	MSF, CE	2,3
• contribute to the maintenance of departmental genetic register systems	MSF, CE	2,3
Be able to explain the benefits and consequences of screening programmes	CE	1,2,3
Behaviours		
Appreciate ethical and social dimensions to population screening	MSF	1,2,4
Understand the central role of patient education	MSF, PS	1,2,4
Appreciate the value of specialised clinics (breast clinics, lipid and cardiovascular risk factor clinics)	CE	2,3,4

15. PATIENT EDUCATION AND DISEASE PREVENTION

Educating Patients about Disease, Investigations and Management

To ensure that the trainee has the knowledge, skills and attitudes to be able to educate patients effectively about genetic disease.

Knowledge	Assessment Methods	GMP
Understand the genetic factors which influence the incidence and prevalence of common conditions	CbD, mini-CEX	1
Understand the factors which influence health and illness – psychological, biological, social, cultural and economic especially poverty	CbD, mini-CEX	1
Understand the influence of lifestyle on health and the factors that influence an individual to change their lifestyle	CbD, mini-CEX	1
Understand the purpose of screening programmes and know in outline the common programmes available within the UK	CbD, mini-CEX	1
Understand the positive and negative effects of screening on the individual	CbD, mini-CEX	1
Demonstrate in practice an appropriate knowledge of the influences of environment and behaviour on health including major factors such as poverty and poor housing, as well as those that might be overlooked	CbD	1
Skills		
Identify opportunities to promote changes in lifestyle and other actions which will positively improve health and/or disease outcomes.	CbD, mini-CEX	1,2
Identify the interaction between mental, physical and social wellbeing in relation to health	CbD, mini-CEX	1
Counsel patients appropriately on the benefits and risks of screening and health promotion activities	CbD, mini-CEX, PS	1,3
Identify patient's ideas, concerns and health beliefs regarding screening and health promotions programmes and be capable of appropriately responding to these	CbD, mini-CEX	1,3
Behaviours		
Encourage patients to access further information and patient support groups	CbD	1,3
Engage in effective team-working around the improvement of health	CbD, MSF	1,3
Encourage where appropriate screening to facilitate early intervention	CbD	1,2,3

Managing Long-Term Conditions and Promoting Patient Self-Care

Work with patients and use their expertise to manage their condition collaboratively and in partnership, with mutual benefit

To pursue a holistic and long term approach to the planning and implementation of patient care, in particular to identify and facilitate the patient's role in their own care

Knowledge	Assessment Methods	GMP
Describe the natural history of diseases and illnesses that run a chronic course	CbD, mini-CEX	1
Define the role of rehabilitation services and the multi-disciplinary team to facilitate long-term care	CbD, mini-CEX	1
Outline the concept of quality of life and how this can be measured whilst understanding the limitations of such measures for individual patients	CbD	1
Outline the concept of patient self-care and the role of the expert patient	CbD, mini-CEX	1
Know, understand and be able to compare and contrast the medical and social models of disability	CbD	1
Understand the relationship between local health, educational and social service provision including the voluntary sector.	CbD	1
Skills		
Develop and agree a management plan with the patient (and carers), ensuring comprehension to maximise self-care within care pathways where relevant	CbD, mini-CEX	1,3
Develop and sustain supportive relationships with patients with whom care will be prolonged and potentially life long	CbD, mini-CEX	1,4
Promote and encourage involvement of patients in appropriate support networks, both to receive support and to give support to others	CbD, PS	1,3
Encourage and support patients in accessing appropriate information	CbD, PS	1,3
Behaviours		
Put patients in touch with the relevant agency including the voluntary sector from where they can procure items and other help as appropriate	CbD, mini-CEX	1,3
Show willingness to maintain a close working relationship with other members of the multi-disciplinary team, primary and community care	CbD, mini-CEX, MSF	3
Recognise and respect the role of family, friends and carers in the management of the patient with a long term condition and the effect of full time caring on carer well-being	CbD, mini-CEX, PS	1,3

16. WORKING WITH COLLEAGUES

Interactions Between:

Hospital & GP

Hospital & Other Agencies e.g. Social Services

Medical and Surgical Specialties

To demonstrate good working relationships with Colleagues		
Knowledge	**Assessment Methods**	**GMP**
Know the roles and responsibilities of team members and know how a team works effectively	CbD, MSF	1,2
Know the role of multidisciplinary management in genetic disorders.	CbD, MSF	1
The principles of effective inter-professional collaboration to optimise patient, or population, care	CbD, MSF	1
Demonstrate knowledge of facilitation and conflict resolution methods	MSF	1,2,4
Skills		
Show leadership, delegate and supervise safely	CbD, MSF	1,2
Be able to communicate effectively	MSF	1,2,3
Recognise when input from another specialty is required for individual patients	MSF	1,2
Be able to work effectively with GPs, other medical and surgical specialists and other health care professionals	MSF	1,2
Employ behavioural management skills with colleagues to prevent and resolve conflict and enhance collaboration	CbD, mini-CEX, MSF	1,3
Demonstrate the ability to facilitate, chair, and contribute to meetings	MSF	1,3
Prepare for meetings - reading agendas, understanding minutes, action points and background research on agenda items	MSF	1,3
Maintain and routinely practice critical self-awareness, including able to discuss strengths and weaknesses with supervisor, recognising external influences and changing behaviour accordingly	MSF	1,3
Create open and non-discriminatory professional working relationships with colleagues awareness of the need to prevent bullying and harassment	MSF	1,3
Develop effective working relationships with colleagues and other staff through good communication skills , building rapport and articulating own view	MSF	1,3
Communicate effectively in the resolution of conflicts, providing feedback, and identifying and rectifying team dysfunction	MSF	1,3
Behaviours		
Foster a supportive and respectful environment where there is open and transparent communication between all team members	CbD, mini-CEX, MSF	1,3
Ensure appropriate confidentiality is maintained during communication with any member of the team	CbD, mini-CEX, MSF	1,3

17. TEACHING AND EDUCATIONAL SUPERVISION

To Have the Skills, Attitudes and Practices of a Competent Teacher

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).

Knowledge	Assessment Methods	GMP
Outline the structure of an effective appraisal interview	CbD	1
Differentiate between formative and summative assessment and define their role in medical education	CbD	1
Outline the role of workplace-based assessments, the assessment tools in use, their relationship to course learning outcomes, the factors that influence their selection and the need for monitoring evaluation	CbD	1
Outline the appropriate local course of action to assist a trainee experiencing difficulty in making progress within their training programme	CbD	1
Skills		
Be able to critically evaluate relevant educational literature	CbD	1
Vary teaching format and stimulus, appropriate to situation and subject	CbD, TO	1
Provide effective feedback and appropriate after teaching, and promote learner reflection	CbD, MSF, TO	1
Conduct developmental conversations as appropriate e.g.: appraisal, supervision, mentoring	CbD, MSF	1
Demonstrate effective lecture, presentation, small group and bed side teaching sessions	CbD, MSF, TO	1,3
Participate in strategies aimed at improving patient education e.g. talking and listening at support group meetings	CbD, MSF, TO	1
Be able to lead departmental teaching programmes including journal clubs	CbD, TO	1
Behaviours		
In discharging educational duties acts to maintain the dignity and safety of patients at all times	CbD, MSF	1,4
Recognise the importance of the role of the physician as an educator within the multi-professional healthcare team and uses medical education to enhance the care of patients	CbD, MSF	1
Encourage discussions with colleagues in clinical settings to colleagues to share knowledge and understanding	CbD, MSF	1,3
Maintain honesty and objectivity during appraisal and assessment	CbD, MSF	1
Show willingness to participate in workplace-based assessments and demonstrates a clear understanding of their purpose	CbD, MSF	1
Demonstrates a willingness to advance own educational capability through continuous learning	CbD, MSF	1
Acts to enhance and improve educational provision through evaluation of own practice	CbD, MSF	1

18. RESEARCH

To Be Able to Plan and Analyse Research

Trainees who wish to acquire extensive research competencies, in addition to those specified in the generic element of the curriculum may undertake a research project as an ideal way of obtaining those competencies; all options can be considered including taking time out of programme to complete a specified project or research degree. Time out of programme needs prospective approval from the SAC and the support of the Postgraduate Dean. Funding will need to be identified for the duration of the research period. A maximum period of 3 years out of programme is allowed.

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.

Knowledge	Assessment Methods	GMP
Know how to use appropriate statistical methods	CE	1
Know the principles of gaining regulatory approvals for clinical research (Ethics, R and D approval, MHRA approval)	CE	1,2
Know how to analyse a scientific paper	CE	1
Outline the GMC guidance on good practice in research	CbD	1
Understand the principles of research governance Outline the differences between audit and research	AA, CbD, mini-CEX	1
Describe how clinical guidelines are produced	CbD	1
Demonstrate a knowledge of research principles	CbD, mini-CEX	1
Outline the principles of formulating a research question and designing a project	CbD, mini-CEX	1
Comprehend principal qualitative, quantitative, bio-statistical and epidemiological research methods	CbD	1
Skills		
Undertake systematic critical review of scientific literature	CbD	1
Ability to frame questions to be answered by a research project	mini-CEX	1
Develop protocols and methods for research	CbD	1
Participate in collaborative research with clinical/scientific colleagues	MSF	1
Be able to accurately analyse data	CE	1
Write and submit a case report or scientific paper	CbD	1
Develop critical appraisal skills and apply these when reading literature	CbD, MSF	1
Behaviours		
Demonstrate curiosity and a critical spirit of enquiry	MSF	1
Humility and the acknowledgement of the contribution of others	MSF	1,3
Follow guidelines on ethical conduct in research and consent for research	CbD	1,2

19. CLINICAL GOVERNANCE

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

The organisational framework for Clinical Governance at local, health authority and national levels.

Understanding of the benefits a patient might reasonably expect from Clinical Governance.

Creating an environment where mistakes and mismanagement of patients can be openly discussed and learned from.

Knowledge	Assessment Methods	GMP
Know about quality improvement methodologies including a range of methods of obtaining feedback from patients, the public, and staff	CbD	1,2
Know the principles and processes of evaluation, audit, research and development, clinical guidelines and standard setting in improving quality	CbD	1,2
Outline a variety of methodologies for developing creative solutions to improving services	CbD	1,2
Skills		
Be an active partaker in clinical governance	MSF	1,2
Assess and analyse situations, services and facilities in order to minimise risk to patients and the public	CbD	1,2
Behaviours		
Act as an advocate for the service	MSF	1,2
Actively seek advice / assistance whenever concerned about patient safety	MSF	1,2
Willing to take responsibility for clinical governance activities, risk management and audit in order to improve the quality of the service	MSF	1,2

Evidence-Based Medicine

Knowledge	Assessment Methods	GMP
Know & understand the principles of evidence-based medicine	CbD, CE	1
Know & understand the types of evidence	CbD, CE	
Understands of the application of statistics in scientific medical practice	CbD, MRCP Part 1	1
Understand the advantages and disadvantages of different study methodologies (randomised control trials, case controlled cohort etc.)	CbD, MRCP Part 1	1
Understand the principles of critical appraisal	CbD	1
Understand levels of evidence and quality of evidence	CbD	1
Understand the role and limitations of evidence in the development of clinical guidelines and protocols	CbD, MRCP Part 1	1
Understand the advantages and disadvantages of guidelines and protocols	CbD	1
Understand the processes that result in nationally applicable guidelines (e.g. NICE and SIGN)	CbD	1

Skills		
Able to critically appraise evidence	CbD	1
Ability to be competent in the use of databases, libraries and the internet	CbD	1
Able to discuss the relevance of evidence with individual patient.	CbD, mini-CEX, MSF	1
Ability to search the medical literature including use of PubMed, Medline, Cochrane reviews and the internet	CbD	1
Appraise retrieved evidence to address a clinical question	CbD	1
Apply conclusions from critical appraisal into clinical care	CbD	1
Identify the limitations of research	CbD	1
Contribute to the construction, review and updating of local (and national) guidelines of good practice using the principles of evidence based medicine	CbD	1
Behaviours		
Display a keenness to use evidence in the support of patient care and own decisions therein.	CbD, mini-CEX	
Keep up to date with national reviews and guidelines of practice (e.g. NICE and SIGN)	CbD	1
Aim for best clinical practice (clinical effectiveness) at all times, responding to evidence-based medicine	CbD, mini-CEX	1
Recognise the occasional need to practise outside clinical guidelines	CbD, mini-CEX	1

Audit

Knowledge	Assessment Methods	GMP
Understand the different methods of obtaining data for audit including patient feedback questionnaires, hospital sources and national reference data	AA, CbD	1
Understand the role of audit (improving patient care and services, risk management etc.)	AA, CbD	1
Understand the steps involved in completing the audit cycle	AA, CbD	1
Understands the working and uses of national and local databases used for audit such as specialty data collection systems, cancer registries, etc. The working and uses of local and national systems available for reporting and learning from clinical incidents and near misses in the UK	AA, CbD	1
Skills		
Involvement in on-going audit.		
Undertake at least one audit project.		
Design, implement and complete audit cycles	AA, CbD	1,2
Contribute to local and national audit projects as appropriate (e.g. NCEPOD, SASM)	AA, CbD	1,2
Support audit by junior medical trainees and within the multi-disciplinary team	AA, CbD	1,2
Behaviours		

Recognise the need for audit in clinical practice to promote standard setting and quality assurance	AA, CbD	1,2

Patient Safety

To understand that patient safety depends on the effective and efficient organisation of care, and health care staff working well together. To understand that patient safety depends on safe systems not just individual competency and safe practice. To understand the risks of treatments and to discuss these honestly and openly with patients so that patients are able to make decisions about risks and treatment options. Ensure that all staff are aware of risks and work together to minimise risk.

To recognise the desirability of monitoring performance, learning from mistakes and adopting no blame culture in order to ensure high standards of care and optimise patient safety

Knowledge	Assessment Methods	GMP
Understand the elements of clinical governance	CbD, MSF	1
Recognise that governance safeguards high standards of care and facilitates the development of improved clinical services	CbD, MSF	1,2
Define local and national significant event reporting systems relevant to specialty	CbD, mini-CEX	1
Recognise importance of evidence-based practice in relation to clinical effectiveness	CbD	1
Outline local health and safety protocols (fire, manual handling etc.)	CbD	1
Understand risk associated with the trainee's specialty work including biohazards and mechanisms to reduce risk	CbD	1
Keep abreast of national patient safety initiatives including National Patient Safety Agency , NCEPOD reports, NICE guidelines etc.	CbD, mini-CEX	1
Understands the investigation of significant events, serious untoward incidents and near misses	CbD, mini-CEX	1
Outline the components of effective collaboration and team working	Cbd	1
Describe the roles and responsibilities of members of the healthcare team	CbD	1
Skills		
Maintain a portfolio of information and evidence, drawn from your medical practice	CbD	2
Reflect regularly on your standards of medical practice in accordance with GMC guidance on licensing and revalidation	AA	1,2,3,4
Practise with attention to the important steps of providing good continuity of care	CbD, mini-CEX	1,3,4
Accurate attributable note-keeping including appropriate use of electronic clinical record systems	CbD, mini-CEX	1,3
Demonstrate leadership and management in the education and training of junior colleagues and other members of the healthcare team	CbD, mini-CEX	1,2,3
Lead and participate in interdisciplinary team meetings	CbD, mini-CEX	3
Provide appropriate supervision to less experienced colleagues	CbD, MSF	3

Behaviours		
Show willingness to participate in safety improvement strategies such as critical incident reporting	CbD, MSF	3
Develop reflection in order to achieve insight into own professional practice	CbD, MSF	3
Demonstrates personal commitment to improve their own performance in the light of feedback and assessment	CbD, MSF	3
Engage with an open no blame culture	CbD, MSF	3
Respond positively to outcomes of audit and quality improvement	CbD, MSF	1,3
Co-operate with changes necessary to improve service quality and safety	CbD, MSF	1,2
Encourage an open environment to foster and explore concerns and issues about the functioning and safety of team working	CbD, MSF	3
Recognise limits of own professional competence and only practise within these.	CbD, MSF	3
Recognise the importance of induction for new members of a team	CbD, MSF	3

20. STRUCTURE OF THE NHS AND THE PRINCIPLES OF MANAGEMENT

Structure of the NHS and the Principles of Management

To display knowledge of the structure and organisation of the NHS nationally and locally.

Knowledge	Assessment Methods	GMP
Understand the local structure of NHS systems in your locality recognising the potential differences between the four countries of the UK	CbD	1
Understand the structure and function of healthcare systems as they apply to your specialty	CbD	1
Understand the consistent debates and changes that occur in the NHS including the political, social, technical, economic, organisational and professional aspects that can impact on provision of service	CbD	1
Demonstrate knowledge of: • The structure, financing, and operation of the NHS and its constituent organisations	MSF	1,2
• Ethical and equality aspects relating to management and leadership e.g. approaches to use of resources/ rationing; approaches to involving the public and patients in decision making	MSF	1,2
• Business management principles: priority setting and basic understanding of how to produce a business plan	MSF	1,2
• The requirements of running of a department, unit or practice relevant to the specialty	MSF	1,2
• Efficient use of clinical resources in order to provide care	MSF	1,2
• Commissioning, funding and contracting arrangements relevant to the specialty	MSF	1,2
• How financial pressures experienced by the specialty department and organisation are managed	MSF	1,2
• Relevant legislation (e.g. Equality and Diversity, Health and Safety, Employment Law) and local Human Resource policies	MSF	1,2
• The duties, rights and responsibilities of an employer, and of a co-worker (e.g. looking after occupational safety of fellow staff)	MSF	1,2
• Individual performance review purpose, techniques and processes, including difference between appraisal, assessment and revalidation	MSF	1,2
• The responsibilities of the various Executive Board members and Clinical Directors or leaders	MSF	1,2
Demonstrate knowledge of organisational performance management techniques and processes	CbD	1
Skills		
Develop skills in managing change and managing people.	MSF	1
Develop leadership skills to play a leading role in developing regional genetic services.	MSF	1

Develop interviewing techniques and those required for performance reviews.	MSF	1
Participate in managerial meetings	CbD	1
Take an active role in promoting the best use of healthcare resources	CbD, mini-CEX	1
Work with stakeholders to create and sustain a patient-centred service	CbD, mini-CEX	1
Employ new technologies appropriately, including information technology	CbD, mini-CEX	1
Demonstrate the ability to develop protocols & guidelines and implementation of these	CbD	1,2
Analyse feedback and comments and, integrate them into plans for the service	CbD	1,2
Use clinical audit with the purpose of highlighting resources required	CbD	1,2
Identify trends, future options and strategy relevant to the specialty and delivering patient services	CbD	1,2
Compare and benchmark healthcare services	CbD	1,2
Behaviours		
Recognise the importance of equitable allocation of healthcare resources and of commissioning	CbD	1,2
Recognise the role of doctors as active participants in healthcare systems	CbD, mini-CEX	1,2
Respond appropriately to health service objectives and targets and take part in the development of services	CbD, mini-CEX	1,2
Recognise the role of patients and carers as active participants in healthcare systems and service planning	CbD, mini-CEX, PS	1,2,3
Show willingness to improve managerial skills (e.g. management courses) and engage in management of the service	CbD, MSF	1
Demonstrate: • Being prepared to accept responsibility	CbD, MSF	1
• Showing commitment to continuing professional development which involves seeking training and self development opportunities, learning from colleagues and accepting constructive criticism	CbD, MSF	1
• Commitment to the proper use of public money. Showing a commitment to taking action when resources are not used efficiently or effectively	CbD, MSF	1

21. INFORMATION TECHNOLOGY, COMPUTER ASSISTED LEARNING AND INFORMATION MANAGEMENT

To Demonstrate Good Use of Information Technology for Patient Care and For Own Personal Development

Demonstrate competence in the use and management of health information.

Knowledge	Assessment Methods	GMP
Know how to retrieve and utilize data recorded in clinical systems.	CbD	1,2
Understanding the range of possible uses for clinical data and information and appreciate the dangers and benefits of aggregating clinical data.	CbD	1,2
Skills		
Demonstrate competent use of database, word processing and statistics programmes	CbD	1
Undertake effective literature searches	CbD	1
Access genetic web sites and specialist databases to undertake searches	CbD, mini-CEX	1
Produce effective computer assisted presentations	CbD, MSF	1
Behaviours		
Be willing to offer advice to lay person on access to appropriate internet sources and support groups	CbD, mini-CEX	1,2
Adopt proactive and enquiring attitude to new technology	CbD	1
Contribute to the development of sensitive validation frameworks to enable patients and their families to make judgements between different sources of information, advice and support	CbD	1,2

4 Learning and Teaching

4.1 The Training Programme

The organisation and delivery of postgraduate training is the statutory responsibility of the General Medical Council (GMC) which devolves responsibility for the local organisation and delivery of training to the deaneries. Each deanery oversees a "School of Medicine" which is comprised of the regional Specialty Training Committees (STCs) in each medical specialty. Responsibility for the organisation and delivery of specialty training in Clinical Genetics in each deanery is, therefore, the remit of the regional Clinical Genetics STC. Each STC has a Training Programme Director who coordinates the training programme in the specialty.

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.

There is flexibility in the delivery of the Clinical Genetics curriculum. Training extends over four years, during which the trainee will be expected to achieve competencies in the following speciality areas: neurogenetics, dysmorphology and foetal medicine, cancer genetics and cardiac genetics.

The sequence of training should ensure appropriate progression in experience and responsibility. The training to be provided at each training site is defined to ensure that, during the programme, the entire curriculum is covered and also that unnecessary duplication and educationally unrewarding experiences are minimised. However, the sequence of training should ideally be flexible enough to allow the trainee to develop a special interest.

Depending on the resources of the training department, either a modular approach will be adopted to facilitate training in the various aspects of Clinical Genetics, or a year by year training approach to cover all the aspects of the curriculum. Progression through the training programme is dependent on documented satisfactory progress submitted to the ARCP review process.

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.

Acting up as a consultant (AUC)

"Acting up" provides doctors in training coming towards the end of their training with the experience of navigating the transition from junior doctor to consultant while maintaining an element of supervision.

Although acting up often fulfills a genuine service requirement, it is not the same as being a locum consultant. Doctors in training acting up will be carrying out a consultant's tasks but with the understanding that they will have a named supervisor at the hosting hospital and that the designated supervisor will always be available for support, including out of hours or during on-call work. Doctors in training will need to follow the rules laid down by the Deanery / LETB within which they work and also follow the JRCPTB rules which can be found at www.jrcptb.org.uk/trainingandcert/Pages/Out-of-Programme.

4.2 Teaching and Learning Methods

The curriculum will be delivered through a variety of learning experiences. Trainees will learn from practice, clinical skills appropriate to their level of training and to their attachment within the department.

Trainees will achieve the competencies described in the curriculum through a variety of learning methods. There will be a balance of different modes of learning from formal teaching programmes to experiential learning 'on the job'. The proportion of time allocated to different learning methods may vary depending on the nature of the attachment within a rotation.

Learning with Peers - There are many opportunities for trainees to learn with other clinical genetic trainees. Supra regional alliances allow study days to be organised at specific times of the year specifically for trainees from all of the centres to capitalise on specialist educational input from the regional genetic centres involved. An active Clinical Genetics trainee network exists on the Internet for the sharing of information.

Examination preparation encourages the formation of self-help groups and learning sets.

Work-based Experiential Learning - The content of work-based experiential learning is decided by the local faculty for education but includes active participation in:

- Out-patient clinics and ward referrals. After initial induction, trainees will see patients in outpatient clinics and wards in both the regional centre and district general hospital outreach clinics and wards, supervised by experienced trained clinicians and genetic counsellors. The degree of responsibility taken by the trainee will increase as competency increases. As experience and clinical competence increase trainees will assess 'new' and 'review' patients and present their findings to their clinical supervisor
- Multi-disciplinary team meetings. There are many situations where clinical problems are discussed with clinicians in other disciplines. These provide excellent opportunities for observation of and participation in clinical reasoning.
- Reflective practice. Opportunities exist for the trainee to review those patients and families seen, encouraging reflective practice. Co-counselling with experienced genetic counsellors enables constructive feedback on the counselling sessions and the development of appropriate professional behaviours in dealing with patients and families. Trainees are encouraged to accompany genetic counsellors at initial contact with a newly referred family to learn how to gather initial background information. The trainee observes and undertakes the whole process of genetic counselling from initial contact to follow-up.

Concentrated Practice in Skills and Procedures - Understanding the methodologies undertaken in the genetics laboratory is essential for the clinical

geneticist who is required to request investigations appropriately and explain and interpret the results of tests for patients and their families as well as fellow professionals.

This understanding is achieved through hands on experience in the laboratories. During specialty training each trainee will spend time in both cytogenetic and molecular genetics laboratories learning the basic procedures and methodologies employed. Apart from laboratory bench experience there will be opportunities for small group tutorials with laboratory scientists.

Many genetic disorders are rare and short blocks of attendance and observation in specialised clinics allows concentrated experience. Examples are:

- Specialist clinics within the Clinical Genetics service based on the specialist expertise of the educational supervisor
- Multi-disciplinary clinics (joint clinics with other specialists)
- Clinics held by non-genetic specialists in areas that impact on clinical genetic practice (e.g. fetal and reproductive medicine)
- Clinics held by non-genetic specialists that allow a greater understanding of the clinical management of conditions, both common (e.g. oncology clinics) or rare disorders (e.g. inherited metabolic disorders) and the management of genetic disorders in non-genetic settings (e.g. haematology clinics)

Formal Postgraduate Teaching – The content of these sessions are determined by the local faculty of medical education and will be based on the curriculum. There are many opportunities throughout the year for formal teaching in the local postgraduate teaching sessions and at regional, national and international meetings. Many of these are organised by the Royal Colleges of Physicians.

Suggested activities include:

- Case presentations
- Journal clubs
- Research and audit projects
- Lectures and small group teaching
- Grand Rounds
- Clinical skills demonstrations and teaching
- Critical appraisal and evidence based medicine and journal clubs
- Joint specialty meetings
- Attendance at training programmes organised on a deanery or regional basis, which are designed to cover aspects of the training programme outlined in this curriculum.

Independent Self-Directed Learning -Trainees will use this time in a variety of ways depending upon their stage of learning. Suggested activities include:

- Reading, including web-based material
- Maintenance of personal portfolio (self-assessment, reflective learning, personal development plan)
- Audit and research projects
- Reading journals
- Achieving personal learning goals beyond the essential, core curriculum

Formal Study Courses - Time to be made available for formal courses is encouraged, subject to local conditions of service. Examples include management courses and communication courses.

External Learning Opportunities

- Attendance at regional and national meetings e.g. Dysmorphology Club, Cancer Genetics Group relevant to the current component of training being undertaken.
- Attendance and presentation at national conferences e.g. Clinical Genetics Society and the British Society of Human Genetics
- Attendance and presentation at international conferences
- Attendance at trainee national courses e.g. genetic counselling
- Participation in the work of patient support groups (medical advisor, committee member)
- After completion their first year of training, trainees may wish to arrange a short period (up to 4 weeks) of overseas experience. Trainees are encouraged to consider visiting a recognised genetic centre in a developing country where there are existing links with local clinicians. However experience in a developed country will also be considered.

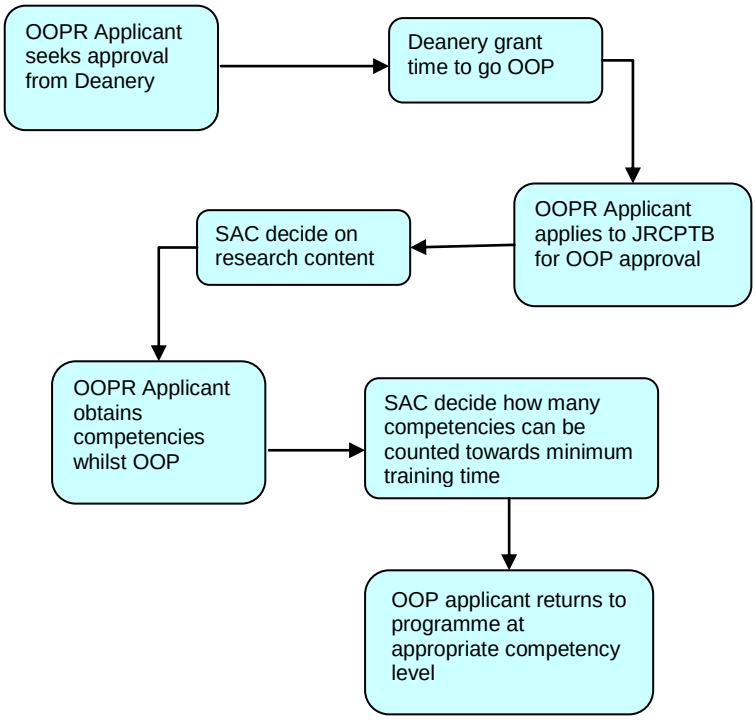
These short electives will require Out Of Programme Experience approval from the deanery and SAC. Although there will be no requirement for formal assessment during the visit the experience gained, mainly through observation and discussion, will inform reflective learning experiences or case based discussions and can be recorded in the ePortfolio. The overseas training period may only be arranged with the agreement of the local Educational Supervisor, Training Consultant and Training Programme Director.

4.3 Research

Trainees, who wish to acquire research competencies, in addition to those specified in their specialty curriculum, may undertake a research project as an ideal way of obtaining those competencies. For those in specialty training, one option to be considered is that of taking time out of programme to complete a specified project or research degree. Applications to research bodies, the deanery (via an OOPR form) and the JRCPTB (via a Research Application Form) are necessary steps, which are the responsibility of the trainee. The JRCPTB Research Application Form can be accessed via the JRCPTB website. It requires an estimate of the competencies that will be achieved and, once completed, it should be returned to JRCPTB together with a job description and an up to date CV. The JRCPTB will submit applications to the relevant SACs for review of the research content including an indicative assessment of the amount of clinical credit (competence acquisition) which might be achieved. This is likely to be influenced by the nature of the research (e.g. entirely laboratory-based or strong clinical commitment), as well as duration (e.g. 12 month Masters, 2-year MD, 3-Year PhD). On approval by the SAC, the JRCPTB will advise the trainee and the deanery of the decision. The deanery will make an application to the GMC for approval of the out of programme research. All applications for out of programme research must be prospectively approved.

Upon completion of the research period the competencies achieved will be agreed by the OOP Supervisor, Educational Supervisor and communicated to the SAC, accessing the facilities available on the JRCPTB ePortfolio. The competencies achieved will determine the trainee's position on return to programme; for example if an ST3 trainee obtains all ST4 competencies then 12 months will be recognised towards the minimum training time and the trainee will return to the programme at ST5. This would be corroborated by the subsequent ARCP.

This process is shown in the diagram below:



Funding will need to be identified for the duration of the research period. Trainees need not count research experience or its clinical component towards a CCT programme but must decide whether or not they wish it to be counted on application to the deanery and the JRCPTB.

A maximum period of 3 years out of programme is allowed and the SACs will recognise up to 12 months towards the minimum training times.

4.4 Academic Training

One training route for clinicians who know from the outset of their training that they wish to become clinical academics is the Academic Clinical Fellowship (ACF) / Academic Clinical Lectureship (ACL) path. ACFs should have both an Academic and a Clinical Educational Supervisor. Academic Clinical Fellowships are three year fixed posts in which 25% of the time is allocated to academic training. Typically this is used to develop a research interest and submit an application for a Clinical Training Fellowship. On completing a Clinical Training Fellowship, the trainee may move on to an Intermediate Training Fellowship or an Academic Clinical Lectureship or a Specialty Registrar position. Academic Clinical Lecturer Posts have a maximum four year duration. A programme is agreed with the Educational Supervisor such that clinical training is completed whilst in the post, and the post ends with Completion of Clinical Training. Typically 50% of time is allocated to academic work, either research or education. SpRs may also apply for Clinical Training Fellowships or other research funding. ACL posts are open to those with an MD/PhD or those who graduated from an MB/PhD programme who meet the entry requirements for specialist training. All applications for research must be prospectively approved by the SAC and the regulator, see www.jrcptb.org.uk for details of the process.

Academic integrated pathways to CCT are a) considered fulltime CCTs as the default position and b) are run through in nature. The academic programmes are CCT programmes and the time set for the CCT is the time set for academic trainees. If a trainee fails to achieve all the required competencies within the notional time period

for the programme, this would be considered at the ARCP, and recommendations to allow completion of clinical training would be made (assuming other progress to be satisfactory) see the guidelines for monitoring training and progress http://www.academicmedicine.ac.uk/careersacademicmedicine.aspx. Extension of a CCT date will be in proportion depending upon the nature of the research and will ensure full capture of the specialty outcomes set down by the Royal College and approved by GMC.

Academic trainees are encouraged to identify an academic mentor, who will not usually be their research supervisor and will often be from outside their geographical area. The Academy of Medical Sciences organises one such scheme (see http://www.acmedsci.ac.uk/index.php?pid=91) but there are others and inclusion in an organised scheme is not a pre-requisite. The Medical Research Society organises annual meetings for clinician scientists in training (see http://www.medres.org.uk/j/index.php?option=com_content&task=view&id=54&Itemid=1) and this type of meeting provides an excellent setting for trainees to meet colleagues and share experiences.

5 Assessment

5.1 The Assessment System

The purpose of the assessment system is to:

- enhance learning by providing formative assessment, enabling trainees to receive immediate feedback, measure their own performance and identify areas for development;
- drive learning and enhance the training process by making it clear what is required of trainees and motivating them to ensure they receive suitable training and experience;
- provide robust, summative evidence that trainees are meeting the curriculum standards during the training programme;
- ensure trainees are acquiring competencies within the domains of Good Medical Practice*;*
- assess trainees' actual performance in the workplace;
- ensure that trainees possess the essential underlying knowledge required for their specialty;
- inform the Annual Review of Competence Progression (ARCP), identifying any requirements for targeted or additional training where necessary and facilitating decisions regarding progression through the training programme;
- identify trainees who should be advised to consider changes of career direction.

The integrated assessment system comprises both workplace-based assessments and knowledge – base assessments. Individual assessment methods are described in more detail below.

Workplace-based assessments will take place throughout the training programme to allow trainees to continually gather evidence of learning and to provide trainees with formative feedback. They are not individually summative but overall outcomes from a number of such assessments provide evidence for summative decision making. The number and range of these will ensure a reliable assessment of the training relevant to their stage of training and achieve coverage of the curriculum.

5.2 Assessment Blueprint

In the syllabus (3.3) the "Assessment Methods" shown are those that are appropriate as **possible** methods that could be used to assess each competency. It is not expected that all competencies will be assessed and that where they are assessed not every method will be used.

5.3 Assessment Methods

The following assessment methods are used in the integrated assessment system:

Examinations and Certificates

- The Certificate Examination of Clinical Genetics (CE)

Information about MRCP (UK), including guidance for candidates, is available on the MRCP(UK) website www.mrcpuk.org

The SAC, Clinical Genetics Society and the Genetic Interest Group are developing a Certificate Examination with the Royal College of Pathologists who currently offer a similar examination for clinical Scientists working in Genetic specialties. We are awaiting formal approval from RCPath and JRCPTB.

This knowledge-based assessment is designed to provide trainees in Clinical Genetics with an assessment of their knowledge of the scientific basis of their discipline. The assessment is a short answer question format based on the successful format used for a similar purpose of testing scientific knowledge in Clinical Biochemistry, and now used in several other scientific disciplines. The assessment will be blueprinted to the Clinical Genetics curriculum. The questions will be set by experts in the field from the Royal Colleges of Physicians and Pathologists. The examination will be run by the College of Pathologists which has been running examinations for laboratory geneticists for many years.

Workplace-Based Assessments WPBAs

- Multi-Source Feedback (MSF)
- mini-Clinical Evaluation Exercise (mini-CEX)
- Case-Based Discussion (CbD)
- Patient Survey (PS)
- Audit Assessment (AA)
- Teaching Observation (TO)

These methods are described briefly below. More information about these methods including guidance for trainees and assessors is available in the ePortfolio and on the JRCPTB website www.jrcptb.org.uk. Workplace-based assessments should be recorded in the ePortfolio. The workplace-based assessment methods include feedback opportunities as an integral part of the process; this is explained in the guidance notes provided for the techniques.

Multisource Feedback (MSF)
This tool is a method of assessing generic skills such as communication, leadership, team working, reliability etc., across the domains of Good Medical Practice. This provides objective systematic collection and feedback of performance data on a trainee, derived from a number of colleagues. 'Raters' are individuals with whom the trainee works, and includes doctors, administration staff, and other allied professionals. The trainee will not see the individual responses by raters, feedback is given to the trainee by the Educational Supervisor.

mini-Clinical Evaluation Exercise (mini-CEX)
This tool evaluates a clinical encounter with a patient to provide an indication of competence in skills essential for good clinical care such as history taking, examination and clinical reasoning. The trainee receives immediate feedback to aid learning. The mini-CEX can be used at any time and in any setting when there is a trainee and patient interaction and an assessor is available.

Case based Discussion (CbD)
The CbD assesses the performance of a trainee in their management of a patient to provide an indication of competence in areas such as clinical reasoning, decision-making and application of medical knowledge in relation to patient care. It also serves as a method to document conversations about, and presentations of, cases by trainees. The CbD should include discussion about a written record (such as written case notes, out-patient letter, and discharge summary). A typical encounter might be when presenting newly referred patients in the out-patient department.

Patient Survey (PS)
Patient Survey addresses issues, including behaviour of the doctor and effectiveness of the consultation, which are important to patients. It is intended to assess the trainee's performance in areas such as interpersonal skills, communication skills and professionalism by concentrating solely on their performance during one consultation.

Audit Assessment Tool (AA)
The Audit Assessment Tool is designed to assess a trainee's competence in completing an audit. The Audit Assessment can be based on review of audit documentation OR on a presentation of the audit at a meeting. If possible the trainee should be assessed on the same audit by more than one assessor.

Teaching Observation (TO)
The Teaching Observation form is designed to provide structured, formative feedback to trainees on their competence at teaching. The Teaching Observation can be based on any instance of formalised teaching by the trainee which has been observed by the assessor. The process should be trainee-led (identifying appropriate teaching sessions and assessors).

5.4 Decisions on Progress (ARCP)

The Annual Review of Competence Progression (ARCP) is the formal method by which a trainee's progression through her/his training programme is monitored and recorded. ARCP is not an assessment – it is the review of evidence of training and assessment. The ARCP process is described in A Reference Guide for Postgraduate Specialty Training in the UK (the "Gold Guide" – available from www.mmc.nhs.uk). Deaneries are responsible for organising and conducting ARCPs. The evidence to be reviewed by ARCP panels should be collected in the trainee's ePortfolio.

The ARCP Decision Aid is included in section 5.5, giving details of the evidence required of trainees for submission to the ARCP panels.

5.5 ARCP Decision Aid

Clinical Genetics Specialist Training

Year of Training	ARCP year 3 (End of ST3)	ARCP year 4 (End of ST4)	ARCP year 5 (End of ST5 = PYA)	ARCP year 6 (End of ST6 = CCT)
Expected competence	Trainees should be competent in the initial assessment of patients presenting with a common genetic disorder. They should be competent in putting forward a basic plan for genetic investigations and clinical management.	Trainees should be competent in the assessment of patients presenting with the majority of common genetic conditions. Trainees should be competent in their approach to the assessment of patients with some rare genetic disorders.	Trainees should be autonomously competent in the assessment and management of patients presenting with common genetic disorders. Trainees should be competent in the assessment and management of genetic disorders presenting acutely (for example in pregnancy).	Trainees should be autonomously competent in the assessment and management of patients presenting with genetic conditions.
Assessments:				
Clinical Genetics Certificate Examination		Attempt/pass CE	Attempt/pass CE	Passed CE in order to obtain CCT
MSF	Satisfactory		Satisfactory	
Patient Survey		Satisfactory		Satisfactory
mini-CEX	4 mini-CEX with emphasis on recording family tree, clinical history or clinical examination of patients with genetic conditions. (the Educational Supervisor should choose the topic to be assessed for at least 1 of the	4 mini-CEX with emphasis on the assessment, management and genetic counselling of patients with genetic conditions. (the Educational Supervisor should choose the topic to be assessed for at least 1 of the	4 mini-CEX with emphasis on the assessment, management and genetic counselling of patients with genetic conditions including those with more complex pedigrees or genetic disorders (the Educational Supervisor	4 mini-CEX on the assessment, management and genetic counselling of patients with genetic conditions with the emphasis on complex disorders. (the Educational Supervisor should choose the topic to be

Year of Training	ARCP year 3 (End of ST3)	ARCP year 4 (End of ST4)	ARCP year 5 (End of ST5 = PYA)	ARCP year 6 (End of ST6 = CCT)
	mini-CEX)	mini-CEX)	should choose the topic to be assessed for at least 1 of the mini-CEX)	assessed for at least 1 of the mini-CEX)
CBD	4 CBD with emphasis on family tree, clinical history or clinical findings in patients with genetic conditions. (the Educational Supervisor should choose the topic to be assessed for at least 1 of the CbD)	4 CBD with emphasis on the assessment, management and genetic counselling of patients with genetic conditions. (the Educational Supervisor should choose the topic to be assessed for at least 1 of the CbD)	4 CBD with emphasis on the assessment, management and genetic counselling of patients with genetic conditions including those with more complex pedigrees or genetic disorders. (the Educational Supervisor should choose the topic to be assessed for at least 1 of the CbD)	4 CBD on the assessment, management and genetic counselling of patients with genetic conditions with the emphasis on complex disorders (the Educational Supervisor should choose the topic to be assessed for at least 1 of the CbD)
Adult Life Support and Paediatric Life Support	Must have valid ALS and PLS	Must have valid ALS and PLS	Must have valid ALS and PLS	Must have valid ALS and PLS
Audit	Evidence of participation in an audit.	Evidence of participation in an audit.	Evidence of completion of an audit with major involvement in design, implementation, analysis and presentation of results and recommendations.	Satisfactory portfolio of audit involvement.(AA)
Research	Evidence of critical thinking around relevant clinical questions.	Evidence of critical thinking around relevant clinical questions.	Evidence of developing research awareness and competence through participation in research studies, critical reviews, presentation at relevant research meetings or participation in courses.	Satisfactory academic portfolio with evidence of research awareness and competence. Evidence could include a completed study with presentations or publication, a completed higher degree with a research component or a

Year of Training	ARCP year 3 (End of ST3)	ARCP year 4 (End of ST4)	ARCP year 5 (End of ST5 = PYA)	ARCP year 6 (End of ST6 = CCT)
				research degree (MD or PhD).
Teaching	Evidence of participation in teaching of medical students, junior doctors, genetic counsellors or other HPs. Assessed by TO.	Evidence of participation in teaching of medical students, junior doctors, genetic counsellors or other HPs.	Evidence of participation in teaching with results of students' evaluation. Evidence of understanding of the principles of adult education via training course. Assessed by TO.	Portfolio evidence of ongoing evaluated participation in teaching. Evidence of implementation of the principles of adult education.
Management	Evidence of awareness of and participation in some aspect of management systems: examples might include responsibility for organising rotas, teaching sessions or journal clubs.	Evidence of awareness of and participation in some aspect of management systems: examples might include responsibility for organising rotas, teaching sessions or journal clubs.	Evidence of awareness of managerial structures and functions within the NHS: this could include attendance at relevant courses or participation in relevant local management meetings with defined responsibilities.	Evidence of understanding of managerial structures: for example reflective portfolio entries regarding relevant NHS management activities.
Events giving concern	The following events occurring at any time may trigger review of trainee's progress and possible targeted training: issues of professional behaviour; poor performance in work-place based assessments, poor MSF performance; issues arising from supervisor report or issues of governance including patient safety.			

5.6 Penultimate Year Assessment (PYA)

The penultimate ARCP prior to the anticipated CCT date will include an external assessor from outside the training programme. JRCPTB and the deanery will coordinate the appointment of this assessor. This is known as “PYA”. Whilst the ARCP will be a review of evidence, the PYA will include a face to face component.

5.7 Complaints and Appeals

All workplace-based assessment methods incorporate direct feedback from the assessor to the trainee and the opportunity to discuss the outcome. If a trainee has a complaint about the outcome from a specific assessment this is their first opportunity to raise it.

Appeals against decisions concerning in-year assessments will be handled at deanery level and deaneries are responsible for setting up and reviewing suitable processes. If a formal complaint about assessment is to be pursued this should be referred in the first instance to the chair of the Specialty Training Committee who is accountable to the regional deanery. Continuing concerns should be referred to the Associate Dean.

6 Managing Curriculum Implementation

The introduction of a structured competency-based training programme for Clinical Genetics and the adoption of competency assessment procedures represent a major departure from the former approach to postgraduate training. Their incorporation in a new legal framework imposes a discipline on all those involved in the educational process. It is essential that there should be an explicit partnership between trainees and those responsible for training, so that trainees receive adequate support and guidance throughout the training period.

In turn there is a new responsibility placed on trainees to evaluate their own strengths and weaknesses and to seek out the educational opportunities that they require to correct any deficiencies.

6.1 Intended Use of Curriculum by Trainers and Trainees

This curriculum is a web-based document which is available from the Joint Royal Colleges of Physicians Training Board (JRCPTB) website www.jrcptb.org.uk for distribution to all participants in Clinical Genetics as appropriate and requested. Hard copies of the curriculum can be prepared at any time from the electronic sources.

The educational supervisors and trainers will be expected to use the curriculum as the basis of their discussion with Trainees. Both trainers and trainees are expected to have a good knowledge of the curriculum and should use it as a guide for their training programme.

Each trainee will engage with the curriculum by maintaining a Training Record (portfolio). The trainee will use the curriculum to develop learning objectives and reflect on learning experiences.

6.2 Trainees: Responsibilities for Curriculum Implementation

One of the basic principles of a competency-based workplace-centred education and training programme is that the trainee is firmly at the centre, not only as the apprentice and raison d’etre for the programme, but as the initiator and responsible

person to ensure that education and training takes place and has a successful outcome. The curriculum for a competency-based programme puts the emphasis on learning rather than teaching.

Whilst specialty advisers and educational bodies can set curricula and lay down standards to be achieved, and educational supervisors and trainers can facilitate the availability of learning opportunities and resources, it is the trainee with the motivation, drive and enthusiasm to undertake specialty training who must ensure that the circumstances are present and appropriate for their full participation, giving them the best chance for a successful and timely outcome.

6.3 Means of Ensuring Curriculum Coverage

The details of how the curriculum is covered in an individual training programme and workplace unit is the responsibility of the Deanery and the Programme Director. The need to show how Clinical Geneticists are progressing in their achievement of learning outcomes has been and will continue to be a strong stimulus to ensure that all curriculum objectives are met.

Clinical Geneticists will provide feedback on their training so that the training programme can be modified as necessary.

6.4 Supervision

The responsibilities of supervisors have been defined by GMC in the document "Operational Guide for the PMETB Quality Framework". These definitions have been agreed with the National Association of Clinical Tutors, the Academy of Medical Royal Colleges and the Gold Guide team at MMC, and are reproduced below:

Clinical Supervisor
A trainer who is selected and appropriately trained to be responsible for overseeing a specified trainee's clinical work and providing constructive feedback during a training placement. Some training schemes appoint an Educational Supervisor for each placement. The roles of Clinical and Educational Supervisor may then be merged.

Educational Supervisor
A trainer who is selected and appropriately trained to be responsible for the overall supervision and management of a specified trainee's educational progress during a training placement or series of placements. The Educational Supervisor is responsible for the trainee's Educational Agreement.

Each trainee has an Educational Supervisor (ES) who must be a consultant clinical geneticist.

The trainee should identify the ES in the preparatory stages of enrolment. The TPD has the responsibility for meeting the nominated ES and approving the choice on behalf of the Postgraduate Dean (PGD.)

Training
The ES must undergo induction and training in the responsibilities, skills and processes of supervision of a trainee in Clinical Genetics.

Further Training
It is expected that ES will undergo refresher training in the role and responsibility of educational supervision.

Additionally, it is appropriate that ES should have the opportunity for additional training in areas of the role appropriate for educational supervision (e.g. appraisal, workplace-based assessment, reflective practice, helping trainees in difficulty). Such programmes are available from the Deanery.

The Educational Supervisor, when meeting with the trainee, should discuss issues of clinical governance, risk management and any report of any untoward clinical incidents involving the trainee. The Educational Supervisor should be part of the clinical specialty team. Thus if the clinical directorate (clinical director) have any concerns about the performance of the trainee, or there were issues of doctor or patient safety, these would be discussed with the Educational Supervisor. These processes, which are integral to trainee development, must not detract from the statutory duty of the trust to deliver effective clinical governance through its management systems.

Opportunities for feedback to trainees about their performance will arise through the use of the workplace-based assessments, regular appraisal meetings with supervisors, other meetings and discussions with supervisors and colleagues, and feedback from ARCP. Clinical Genetics is a multidisciplinary specialty and there will be opportunities for constructive feedback in both formal and informal settings from supervising consultant specialists, genetic counsellors, specialist laboratory staff as well as service users.

6.5 Appraisal

A formal process of appraisals and reviews underpins training. This process ensures adequate supervision during training and provides continuity between posts and different supervisors and is one of the main ways of providing feedback to trainees. All appraisals should be recorded in the ePortfolio

Induction Appraisal

The trainee and educational supervisor should have an appraisal meeting at the beginning of each post to review the trainee's progress so far, agree learning objectives for the post ahead and identify the learning opportunities presented by the post. Reviewing progress through the curriculum will help trainees to compile an effective Personal Development Plan (PDP) of objectives for the upcoming post. This PDP should be agreed during the Induction Appraisal. The trainee and supervisor should also both sign the educational agreement in the ePortfolio at this time, recording their commitment to the training process.

Mid-point Review

This meeting between trainee and educational supervisor is mandatory (except when an attachment is shorter than 6 months), but is encouraged particularly if either the trainee or educational or clinical supervisor has training concerns or the trainee has been set specific targeted training objectives at their ARCP. At this meeting trainees should review their PDP with their supervisor using evidence from the e-portfolio. Workplace-based assessments and progress through the curriculum can be reviewed to ensure trainees are progressing satisfactorily, and attendance at educational events should also be reviewed. The PDP can be amended at this review.

End of Attachment Appraisal

Trainees should review the PDP and curriculum progress with their educational supervisor using evidence from the ePortfolio. Specific concerns may be highlighted from this appraisal. The end of attachment appraisal form should record the areas

where further work is required to overcome any shortcomings. Further evidence of competence in certain areas may be needed, such as planned workplace-based assessments, and this should be recorded. If there are significant concerns following the end of attachment appraisal then the programme director should be informed

6.6 Recording Progress

On enrolling with JRCPTB trainees will be given access to the ePortfolio for Clinical Genetics The ePortfolio allows evidence to be built up to inform decisions on a trainee's progress and provides tools to support trainees' education and development.

The trainee's main responsibilities are to ensure the ePortfolio is kept up to date, arrange assessments and ensure they are recorded, prepare drafts of appraisal forms, maintain their personal development plan, record their reflections on learning and record their progress through the curriculum.

The supervisor's main responsibilities are to use ePortfolio evidence such as outcomes of assessments, reflections and personal development plans to inform appraisal meetings. They are also expected to update the trainee's record of progress through the curriculum, write end-of-attachment appraisals and supervisor's reports.

In addition trainees should keep a log book record of cases seen by date and diagnosis which can be reviewed with their Educational Supervisor at the mid-point review, end of attachment appraisal or ARCP.

7 Curriculum Review and Updating

The specialty curriculum will be reviewed and updated with minor changes on an annual basis. Feedback on the curriculum will be solicited as a standing item on the agenda of the SAC meeting in May which is attended by Specialty Advisers from all Regions. SAC trainee representative members from England, Scotland and Wales have been encouraged to consult their colleagues and to feedback at the May SAC meeting. The Genetic Interest Group is also represented on the SAC. In addition, the curriculum will be subject to three-yearly formal review within the SAC. This will be informed by curriculum evaluation and monitoring. The SAC will have available to it:

- The trainees' survey, which will include questions pertaining to their specialty (GMC to provide)
- Specialty-specific questionnaires
- Reports from other sources such as educational supervisors, programme directors, specialty deans, service providers and patients, and the National Health Service
- Trainee representation on the Deanery STC and the SAC of the JRCPTB
- Informal trainee feedback during appraisal, ARCP, etc.

Evaluation will address:

- The relevance of the learning outcomes to clinical practice
- The balance of work-based and off-the-job learning
- Quality of training in individual posts
- Feasibility and appropriateness of on-the-job assessments in the course of training programmes
- Availability and quality of research opportunities

- Current training affecting the service

Evaluation will be the responsibility of the JRCPTB and GMC. These bodies must approve any significant changes to the curriculum.

Interaction with the NHS will be particularly important to understand the performance of specialists within the NHS and feedback will be required as to the continuing needs for that specialty as defined by the curriculum. It is likely that the NHS will have a view as to the balance between generalist and specialist skills, the development of generic competencies and, looking to the future, the need for additional specialist competencies and curricula. In establishing specialty issues which could have implications for training, the SAC will produce a summary report to discuss with the NHS employers and ensure that conclusions are reflected in curriculum reviews.

In addition to the input to the SAC, trainee contribution to curriculum review will be facilitated through the involvement of trainees in local faculties of education and through informal feedback during appraisal, ARCP, and College meetings.

The SAC will respond rapidly to changes in service delivery. Regular review will ensure the coming together of all the stakeholders needed to deliver an up-to-date, modern specialty curriculum. The curriculum will indicate the last date of formal review monitoring and document revision.

8 Equality and Diversity

The Royal Colleges of Physicians will comply, and ensure compliance, with the requirements of equality and diversity legislation, such as the:

- Race Relations (Amendment) Act 2000
- Disability Discrimination Act 1995
- Human Rights Act 1998
- Employment Equality (Age) Regulation 2006
- Special Educational Needs and Disabilities Act 2001
- Data Protection Acts 1984 and 1998

The Federation of the Royal Colleges of Physicians believes that equality of opportunity is fundamental to the many and varied ways in which individuals become involved with the Colleges, either as members of staff and Officers; as advisers from the medical profession; as members of the Colleges' professional bodies or as doctors in training and examination candidates. Accordingly, it warmly welcomes contributors and applicants from as diverse a population as possible, and actively seeks to recruit people to all its activities regardless of race, religion, ethnic origin, disability, age, gender or sexual orientation.

Deanery quality assurance will ensure that each training programme complies with the equality and diversity standards in postgraduate medical training as set by GMC.

Compliance with anti-discriminatory practice will be assured through:

- monitoring of recruitment processes;
- ensuring all College representatives and Programme Directors have attended appropriate training sessions prior to appointment or within 12 months of taking up post;
- Deaneries must ensure that educational supervisors have had equality and diversity training (at least as an e learning module) every 3 years

- Deaneries must ensure that any specialist participating in trainee interview/appointments committees or processes has had equality and diversity training (at least as an e module) every 3 years.
- ensuring trainees have an appropriate, confidential and supportive route to report examples of inappropriate behaviour of a discriminatory nature. Deaneries and Programme Directors must ensure that on appointment trainees are made aware of the route in which inappropriate or discriminatory behaviour can be reported and supplied with contact names and numbers. Deaneries must also ensure contingency mechanisms are in place if trainees feel unhappy with the response or uncomfortable with the contact individual.
- monitoring of College Examinations;
- ensuring all assessments discriminate on objective and appropriate criteria and do not unfairly disadvantage trainees because of gender, ethnicity, sexual orientation or disability (other than that which would make it impossible to practise safely as a physician). All efforts shall be made to ensure the participation of people with a disability in training.