

### Assessment Blueprint for Clinical Genetics

Curriculum area	Competence	Exam	Mini-CEX	MSF	CBD	Patient Survey	Generic	PFolio
<b>A) History, Examination, Investigations, Management &amp; Note keeping Skills:</b> <b>Learning Objective:</b> To be able to establish genetic diagnoses by means of clinical history taking, physical examination and use of appropriate investigations and to provide clinical genetic management for patients and families.								
	<b>(i) Pre-clinic preparation</b>							
<b>Knowledge</b>	Knowledge of relevant disorder acquired by background reading			X	X			
<b>Skills</b>	Be able to review medical records and identify information sources including databases and literature searches.				X			
<b>Attitudes &amp; Behaviours</b>	Appreciate the importance of identifying key issues and being prepared to deal with these.		X		X			
	<b>(ii) History</b>							
<b>Knowledge</b>	Define the patterns of symptoms found in patients presenting with genetic disease.	X			X			
	Recognise reliable and unreliable family history data and identify sources for verification.		X		X			
<b>Skills</b>	Be able to take and analyse a clinical history in a relevant, succinct and logical manner.		X		X			
	Be able to overcome difficulties of language, physical and mental impairment.		X	X		X		
	Use interpreters and advocates appropriately. Elicit family history information in a sensitive and understanding manner.		x	X		x		
	Draw complex pedigrees accurately, including consanguinity loops, recording appropriate information.		x		x			
<b>A &amp; B</b>	Show empathy with patients.		X	X		X	X	

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	Appreciate the importance of psychological and social factors of patients and relatives in genetic disease.				X			
	Attention to detail and accuracy in collecting and checking family history and medical data.		X		X			
	Appreciate the confidentiality and ethical issues arising from family history gathering				X			
	<b>(iii) Examination</b>							
<b>Knowledge</b>	Define the pathophysiological basis of physical signs.	X			X			
	Define the clinical signs found in genetic diseases.	X			X			
<b>Skills</b>	Be able to perform a reliable and appropriate examination to elicit relevant signs of genetic disease.		X					
	Perform examination appropriately in situations involving cultural sensitivity.		X	X		X		
	Understand when additional specialist examination is required.		X		X			
<b>A&amp;B</b>	Respect patients' dignity and confidentiality. Acknowledge cultural issues.		X	X	X	X	X	
	Appropriately involve relatives.				X		X	
	Appreciate the need for a chaperone.						X	
	<b>(iv) Investigations including imaging</b>							
<b>Knowledge</b>	Know the predictive value of results of investigations.	X			X			
	Define the pathophysiological basis of	X			X			

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	investigations.							
	Define the indications for investigations.	X			X			
	Define the risks and benefits of investigations.	X			X			
	Know the cost effectiveness of individual investigation.				X			
<b>Skills</b>	Ability to prioritise investigations and interpret the results.	X	X		X			
	Ability to perform investigations competently where relevant.				X			
	Ability to liaise and discuss investigations with colleagues and to order them appropriately.			X	X			
<b>A&amp;B</b>	Understand the importance of working with other health care professionals and team working.			X				
	Show a willingness to provide explanation to patient as to rationale for investigations, and possible unwanted effects.		X		X	X		
	Persistence in seeking diagnoses.				X			
	<b>(v)Diagnosis and management</b>							
<b>Knowledge</b>	Recognise pitfalls in single gene inheritance including variable expressivity and reduced penetrance, somatic and gonadal mosaicism.	X			X			
	Be able to formulate differential diagnoses for genetic disorders.	X			X			
<b>Skills</b>	Present genetic information to a patient in a sensitive and understanding manner.		X	X	X	X		
	Calculate genetic risk in single gene disorders by hand	X			X			

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	Calculate genetic risk by use of a computer programme				X			
	Use computerized genetic databases and registers for information retrieval.				X			
	Present undiagnosed cases to colleagues, including dysmorphology club meetings							X
<b>A&amp;B</b>	Show appropriate attitudes towards patients and their symptoms and be conscious of religious or other philosophical contexts particularly in respect to prenatal diagnosis.		X		X			
	Clearly and openly explain management options.		X		X			
	Sensitivity in breaking bad news.		X	X	X	X		
	Appreciate the impact of diagnosing serious genetic conditions on family relationships.				X			
	<b>vi) Note keeping, letters etc</b>							
<b>Knowledge</b>	Use of email, internet and the telephone.						X	
	Define the structure, function and legal implications of medical records & medico-legal reports.						X	
	Know the relevance of the data protection pertaining to patient confidentiality						X	
<b>Skills</b>	Record concisely, accurately, confidentially and legibly the appropriate elements of the history, examination, results of investigations, differential diagnosis and management plan.						X	
	Date and sign all records.						X	

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	Be able to write outpatient letters to professional colleagues.						X	
	Be able to write appropriate summary letters to patients and their families.				X	X		
	Be able to write appropriate letters in response to complaints.						X	
<b>A&amp;B</b>	Appreciate the importance of timely dictation cost effective use of medical secretaries and the growing use of electronic communication.						X	
	Be aware of the need for prompt and accurate communication with primary care and other agencies.						X	
	Show courtesy towards medical secretaries and clerical staff.			X			X	
<b>B) Time Management and Decision Making:</b> <b>Learning Objective:</b> To demonstrate that the trainee has the knowledge, skills and attitudes to manage time and problems effectively.								
	<b>(i) Time management</b>							
<b>Knowledge</b>	Know which patients/tasks take priority.			X			X	
<b>Skills</b>	Start with the most important tasks.						X	
	Work more efficiently as clinical skills develop.						X	
	Recognise when he/she is falling behind and re-prioritise or call for help.			X				
<b>A&amp;B</b>	Have realistic expectations of tasks to be completed by self and others.			X				
	Willingness to consult and work as part of a team.			X				
	<b>(ii) Decision making</b>							

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<b>Knowledge</b>	Understand clinical priorities for investigation and management.			X	X			
<b>Skills</b>	Analyse and manage clinical problems.		X		X			
<b>A&amp;B</b>	Be flexible and willing to change in the light of changing conditions.			X			X	
	Be willing to ask for help.			X			X	
C) Procedures: <b>Learning objective:</b> to demonstrate proficiency in clinical procedures related to genetics.								
	<b>Phlebotomy</b>							
<b>Knowledge</b>	Knowledge of technique						X	
<b>Skills</b>	Ability to take blood samples from adults and children, including those with special needs						X	
<b>A&amp;B</b>	Understand the stress of the technique and obtain consent.						X	
	<b>Hair root extraction</b>							
<b>Knowledge</b>	Knowledge of technique and indications for use		X				X	
<b>Skills</b>	Ability to collect samples suitable for analysis		X				X	
<b>A&amp;B</b>	Explain procedure appropriately and obtain consent		X				X	
	<b>Skin biopsy</b>							
<b>Knowledge</b>	Knowledge of technique and indications for use		X				X	
<b>Skills</b>	Demonstrate ability to obtain samples suitable for analysis		X				X	
<b>A&amp;B</b>	Explain procedure appropriately and obtain consent		X				X	
	<b>Clinical photography</b>							
<b>Knowledge</b>	Knowledge of technique Understand importance and confidentiality of photographic records.						X	

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<b>Skills</b>	Demonstrate ability to take photographs of sufficient quality for clinical use		X					
	Use of digital photography and storage of data						X	
<b>A&amp;B</b>	Explain the need for clinical photography and obtain consent		X				X	
<b>COMMUNICATION SKILLS AND GENETIC COUNSELLING</b>								
<p><b>Learning Objective:</b> 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).</p>								
<b>(i) Within a consultation</b>								
<b>Knowledge</b>	Know how to structure the interview to identify the patient's: <ul style="list-style-type: none"> <li>• concerns/priorities/agenda</li> <li>• expectations</li> <li>• understanding</li> <li>• acceptance</li> </ul>		X					
<b>Skills</b>	Listen.		X					
	Use open questions followed by appropriate closed questions.		X					
	Avoid jargon and use familiar language.		X					
<b>Skills</b>	Be able to communicate both verbally and in writing to patients whose first language may not be English in a manner that they understand.		X	X	X			
	Use interpreters appropriately.				X			
	Communicate effectively with people who have		X		X			

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	hearing or visual disability.							
	Give clear information and feedback to patients and share information with relatives when appropriate		X		X	X		
	Reassure 'worried well' patients, appropriately		X				X	
<b>A&amp;B</b>	Demonstrate an understanding of the need for: <ul style="list-style-type: none"> <li>• involving patients in decisions</li> <li>• offering choices</li> <li>• respecting patients views</li> <li>• dress and appearance to be appropriate to the clinical situation and patient sensibility</li> </ul>		X				X	
<b>(ii) Breaking bad news</b>								
<b>Knowledge</b>	Know how to structure the interview and where it should take place.						X	
	Be aware of the normal bereavement process and behaviour.				X		X	
<b>Skills</b>	Be able to break bad news in steps appropriate to the understanding of the individual and be able to support distress.			X			X	
	Avoid jargon and use familiar language.		X		X		X	
	Encourage questions.		X			X	X	
	Maintain appropriate hope whilst avoiding inappropriate optimism.						X	
<b>A&amp;B</b>	Act with empathy, honesty and sensitivity.			X			X	
<b>(iii) Specific genetic issues</b>								
<b>Knowledge</b>	Knowledge of ethnic difference in the incidence of genetic disease.	X						
	Understanding of cross-cultural issues including consanguinity and arranged marriages.	X	X		X			

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	Understanding of religious beliefs and attitudes to prenatal diagnosis and assisted reproduction techniques.		X		X			
<b>Skills</b>	Appreciation of factors that influence perception of risk.				X			
	Acquisition of “non-directive” counselling skills. Effective use of co-counsellors.		X	X	X	X		
	Communication of genetic information and risk to children and adolescents.		X	X	X			
	Communication with adults and children with learning disability.		X	X	X			
	Recognising which patients will benefit from referral on to psychological services.					X		
<b>A&amp;B</b>	Appreciate patient and family anxieties, both rational and irrational.		X		X			
	Appreciate that every person is influenced by their own culture and beliefs.		X		X			X
	Identify particular ethnic perspectives on genetic diseases.	X	X		X			
<b>A&amp;B</b>	Appreciate the importance of senior figures / elders in patient education.				X			
	Appreciate the importance of genetic nurses and genetic associates.			X	X			
	Cultivate habit of reflection and discussion with colleagues after counselling sessions.			X	X			X
	Readiness to alter practice in light of experience and feed-back.				X			X
	<b>iv) Complaints</b>							
<b>Knowledge</b>	Have awareness of the local complaints procedures.						X	
	Have an awareness of systems of independent review.						X	

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<b>Skills</b>	Manage dissatisfied patients / relatives.			X			X	
	Anticipate potential problems.				X			
<b>A&amp;B</b>	Act with honesty and sensitivity and promptly.			X	X		X	
	Be prepared to accept responsibility.			X	X		X	
	<b>(v) Communication with Colleagues</b>							
<b>Knowledge</b>	Know how to write a problem orientated letter				X			
	Know how to communicate with members of the MDT			X				
	Know when to phone a GP						X	
	Know when to phone a patient at home				X			
<b>Skills</b>	Use appropriate language.			X	X	X	X	
	Select an appropriate communication method.						X	
<b>A&amp;B</b>	Be prompt and respond courteously and fairly			X			X	
<b>FORMAL GENETICS AND BASIC SCIENCES:</b>								
<b>Learning objective:</b> 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)								
<b>Knowledge</b>	The chromosomal basis of heredity (mitosis and meiosis)	X						
	Mechanisms of origin of numerical and structural chromosome abnormalities	X						
	Behaviour of structural chromosome abnormalities at meiosis	X						
	The chemical structure of DNA and replication	X						
	Central dogma of cell biology: transcription and translation	X						
	Modes of inheritance (Mendelian and non Mendelian)	X				X		

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	Risk calculations including combinatorial probability and Bayes Theorem	X			X			
	The clinical embryology and molecular mechanisms of human malformation syndromes	X			X			
	Principles of teratogenesis and pregnancy associated risks	X			X			
	Mechanisms of mutagenesis and estimation of mutation rates	X			X			
	History of genetics	X						
<b>Skills</b>	Recognition of different inheritance patterns in pedigrees				X			
	Pedigree-based calculation of segregation ratios for structural chromosome abnormalities	X			X			
	Empiric risk calculations (occurrence and recurrence risks)	X			X			
	Perform Bayesian risk calculations including linkage-based risk calculations	X						
	Calculate gene frequencies - the Hardy-Weinberg equilibrium and chi square tests of departure	X						
	Apply knowledge to interpret results of chromosome and molecular genetic analysis.	X			X			
<b>A&amp;B</b>	Identification and critical evaluation of information						X	
	Commitment to lifelong self directed learning						X	
	Use primary sources of data				X		X	
	Appreciation the impact of genetic disorders on individuals and families				X			X
	Appreciate potential benefits and harm of new genetic technologies				X			X
	Appreciate public concerns about the application of new genetic technologies				X			X
<b>COMMON GENETIC REFERRALS:</b>								

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

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	neurofibromatosis							
<b>A&amp;B</b>	Appreciate the role of the general practitioner in management of chronic disease				X		X	X
	Appreciate the role of support groups and be willing to provide appropriate information				X	X	X	
	Apply good clinical care and counselling skills (see sections 1 and 2)		X		X			
<b>NEUROGENETICS:</b> <b>Learning objective:</b> To provide the trainee with the skills and knowledge to recognise genetic causes of central and peripheral nervous system dysfunction								
<b>Knowledge</b>	Genetic aspects and clinical presentation of trinucleotide repeat disorders	X			X			
	Classification and molecular basis of common genetic neuromuscular disorders	X			X			
	Basic neuropathology and differential diagnosis of hereditary dementias	X			X			
	Mitochondrial diseases - clinical, biochemical and genetic features	X			X			
	Genetic causes of mental retardation (static and progressive)	X			X			
	Genetic contribution to autism and autistic spectrum disorders	X			X			
	Genetic contribution to psychiatric disease in adults	X			X			
<b>Skills</b>	Recognise family history data that suggest familial neurological disease		X		X			X
	Verify diagnoses from old hospital records				X		X	
	Be able to confirm clinical signs in affected individuals		X		X			
<b>Skills</b>	Be able to draw up a differential diagnosis and institute appropriate genetic testing	X	X		X			

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	Assessment of symptoms and signs in patients at risk of adult-onset neurogenetic disease		X		X			
	Application of protocols for pre-symptomatic diagnosis of Huntington's disease and other neurodegenerative disorders		X		X			X
	Make timely, appropriate referrals to other specialists such as neurologists, psychologists, psychiatrists, speech therapists			X	X			
<b>A &amp; B</b>	Appreciation of family stresses caused by risk or eventuality of neurodegeneration			X	X			X
	Appreciate social problems encountered by adults with mild/moderate learning disability			X	X			X
	Appreciate issues involved in predictive testing			X	X			X
<b>PAEDIATRIC GENETICS AND DYSMORPHOLOGY:</b> <b>Learning objective:</b> To provide the trainee with the skills and knowledge to make syndrome diagnosis in children								
<b>Knowledge</b>	Identify normal developmental milestones and diagnose delayed development	X	X		X		X	
	Explain morphogenesis in terms of deformation, malformation, disruption and dysplasia	X			X			
	Utilise journals and databases used in syndrome identification				X			X
	Have knowledge of common and rarer dysmorphic syndromes	X			X			X
<b>Skills</b>	Be able to take a relevant history, and perform an appropriate examination, obtain illustrative photographs		X		X			
	Have a rational approach to investigation of children with delayed development and/or dysmorphic syndromes		X		X			
	Formulate differential diagnoses of unknown syndromes	X	X		X			

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	Cultivate critical assessment of database information and case reports to identify uncertainty and subjectivity in syndrome diagnosis				X			X
	Be able to provide a diagnostic service within a multidisciplinary clinical team		X	X	X			
	In malformation syndromes, refer patients appropriately to specialist medical and surgical services				X			X
	Present and discuss cases with colleagues				X		X	X
<b>A &amp; B</b>	Recognise importance of clinical judgement, timing, and tact when diagnosing and informing parents of an infant with serious malformation or handicap			X	X	X		X
	Appreciate the emotional reactions of parents following early diagnosis of syndrome or recognition of developmental delay			X	X	X		
	Appreciate the adverse reaction families may experience following retraction of a previous diagnosis			X	X	X		X
<b>A&amp;B</b>	Appreciate the importance of attending Dysmorphology Group Meetings							X
<b>CANCER GENETICS:</b>								
<b>Learning objective:</b> Trainee is able to diagnose rare cancer syndromes and recognise when common cancers are likely to have a single gene basis. The trainee can recommend targeted screening in individuals who are identified as having increased risk. Trainee can coordinate appropriate molecular genetic testing.								
<b>Knowledge</b>	The genetic and environmental factors that affect risk of developing cancer	X						
	Current recommendations concerning tumour	X			X			

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	surveillance in cancer –prone families							
	Knowledge of clinical features of genetic cancer syndromes	X			X			
	Genetic mechanisms in neoplasia: Knudson’s two-hit hypothesis	X						
	Knowledge of molecular basis of cancer genetic syndromes	X						
<b>Skills</b>	Be able to take a relevant history, perform an appropriate examination and undertake risk estimation using a variety of methods		X		X			
	Use of cancer registers and other sources to verify diagnoses				X			
	Use disease registers (e.g. von Hippel Lindau disease) to support follow-up of affected and at-risk patients				X			X
	Assessment of screening protocols for at-risk relatives				X			X
<b>Skills</b>	Identify at-risk patients and relatives who are eligible to participate in trials of cancer prevention strategies				X			
<b>A&amp;B</b>	Demonstrate awareness of the roles primary care and genetic associates play in assessing families where relatives are at risk of developing cancer				X			
	Inform patients about lifestyle factors that affect cancer risk		X		X	X		
	Support general practitioners with the long-term management of selected patients with familial cancer syndromes				X			
	Liase with other specialists as appropriate e.g. for advice about prophylactic mastectomy and work as a member of a multidisciplinary team				X			
	Understand the impact of cancer risk on				X	X		

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	individuals and families							
<b>PRENATAL DIAGNOSIS AND FETAL DYSMORPHOLOGY</b>								
<b>Learning objective:</b> To provide the trainee with the skills and knowledge to undertake genetic assessment of actual and potential problems in the fetus, and provide parents with advice about prognosis and inheritance.								
<b>Knowledge</b>	Through attendances at fetal post-mortem examinations, know the process and limitations of clinical and laboratory diagnostic procedures				X			X
	Have knowledge of RCPATH guidelines on retention and storage of fetal tissues	X						X
	Know the natural history of prenatally diagnosed conditions including autosomal and sex chromosome aneuploidy syndromes	X			X			
<b>Knowledge</b>	Knowledge of the law pertaining to termination of pregnancy for fetal abnormality	X			X			
<b>Skills</b>	Interpret family history data and trace old medical records				X			X
	Perform post-mortem clinical analysis of the fetus (examination, measurements, photography, radiology, tissue sampling and storage for diagnostic studies)		X		X			
	Use syndrome databases in syndrome diagnosis				X			
	Provide genetic advice for women who may undergo prenatal diagnosis		X		X			
	Assess clinical significance of chromosome, DNA, and fetal imaging studies in the context of fetal abnormality or risk thereof		X		X			
	Formulate differential diagnoses and assess prognosis in collaboration with the fetal medicine team		X		X			X
	Perform risk-assessment when pregnancies are				X			X

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	exposed to hazards such as congenital infections, alcohol, ionising irradiation or drugs							
	Sensitive disclosure of abnormal test results or diagnoses in the antenatal period		X	X		X		
<b>A &amp; B</b>	Appreciate the different perspectives on advantages and disadvantages of prenatal diagnosis in each situation				X			
	Non-judgmental appreciation of the ethical and religious dimensions to prenatal diagnosis		X		X			X
	Awareness of the adverse psychological effects of termination of pregnancy for fetal abnormality				X			
<b>LABORATORY GENETICS</b>								
<b>Learning objective:</b> The trainee acquires skills and knowledge to interpret genetic laboratory results within a clinical setting, by completing an attachment in the genetic laboratories								
<b>Knowledge</b>	Techniques for conventional chromosome analysis in different tissues	X						
	Laboratory techniques and application of new cytogenetic tests e.g. FISH/CGH	X						
	Use of ISCN nomenclature	X						
	Molecular genetic techniques in common usage- (DNA extraction, Southern Blotting, PCR, DNA sequencing)	X						
	Application of DNA-based testing for gene mapping, linkage and mutation detection.	X						
	Potential application of new DNA technologies	X						
	Sensitivity and specificity of laboratory tests	X						
	Use of DNA and molecular cytogenetic methods in pre implantation diagnosis	X						
	Investigative approach to biochemical diagnosis of inborn errors of metabolism (via experience gained	X				X		

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	at metabolic disease clinics)							
	The operation of local and national antenatal and newborn, genetic disease screening programmes	X			X			
<b>Skills</b>	Interpretation of clinical consequences of abnormal karyotypes, enzyme deficiencies and molecular test results	X			X			
	Liaise with molecular and cytogenetics scientists in analysis of test results			X				
<b>Skills</b>	Provide advice to laboratory on the wording of reports to referring clinicians			X	X			X
	Genetic risk calculation based on laboratory test results (e.g. MLINK, Bayesian analysis).	X						
<b>A &amp; B</b>	Awareness of the importance of informed consent that arise in relation to storage of DNA samples and cell lines		X		X			X
	Willingness to liaise with colleagues to interpret laboratory results			X				
<b>ORGANISATION AND PROVISION OF GENETICS SERVICES FOR POPULATIONS</b>								
<b>Learning objective:</b> 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.								
<b>Knowledge</b>	The genetic characteristics in different populations, mutant gene frequencies and disease prevalence	X						
	The factors that influence decisions to instigate programmes of population screening for genetic diseases.	X						
	Define sensitivity, specificity, and predictive values of screening tests	X						
	Knowledge of current screening programmes	X						
	Knowledge of appropriate population-based	X						

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	registers.							
<b>Skills</b>	Team-working with database managers, genetic associates and nurse specialists in: - '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) - family based screening for individuals at high risk of developing cancer			X	X			X
	Contribute to the maintenance of departmental genetic register systems			X	X			X
	Be able to explain the benefits and consequences of screening programmes				X			X
<b>A &amp; B</b>	Appreciate ethical and social dimensions to population screening	X			X			
	Understand the central role of patient education				X			
	Appreciate the value of specialised clinics (breast clinics, lipid and cardiovascular risk factor clinics)				X			
	Encourage patients to adopt a healthier lifestyle with specific emphasis on risk factor avoidance and promotion of behaviours that reduce risk of developing disease		X		X	X		X
<b>JOINT SPECIALIST CLINICS</b>								
<b>Learning objective:</b> To equip the trainee with skills and knowledge to provide genetic advice within multidisciplinary clinic settings								
<b>Knowledge</b>	Genetic contribution to multidisciplinary clinics held with other specialists: Child development Vision Hearing	X			X			

### Assessment Blueprint for Clinical Genetics

Curriculum area	Competence	Exam	Mini-CEX	MSF	CBD	Patient Survey	Generic	PFolio
	Endocrine Skeletal dysplasia Neurological Cranio-facial malformation Tumour surveillance							
<b>Skills</b>	Team working skills			X				
	Develop a special interest clinic							X
	Develop skills and liaisons needed to nurture new services, even in settings such as health centres or child development centres, outside of the genetics department				X			X
<b>A &amp; B</b>	Demonstrate appreciation of the role of other professionals and willingness to be flexible in responding to the needs of colleagues and patients			X	X			X
<b>MAINTAINING GOOD MEDICAL PRACTICE</b>								
<b>Learning objective:</b> To inculcate the habit of life long learning								
<b>Knowledge</b>	Define continuing professional development.							X
<b>Skills</b>	Recognise and use learning opportunities.							X
	Use the potential of study leave to keep oneself up to date.							X
<b>A&amp;B</b>	Be: <ul style="list-style-type: none"> <li>• self motivated</li> <li>• eager to learn,</li> </ul>			X	X			
	Show: <ul style="list-style-type: none"> <li>• Willingness to learn from colleagues.</li> <li>• willingness to accept criticism.</li> </ul>			X	X			
<b>MAINTAINING TRUST</b>								
<b>Learning objective:</b> To ensure that the trainee has the knowledge, skills and attitudes to act in a professional manner at all times.								
	<b>(i) Continuity of care</b>							
<b>Knowledge</b>	Understand the relevance of continuity of care.				X		X	
<b>Skills</b>	Ensure satisfactory completion of reasonable tasks			X			X	

### Assessment Blueprint for Clinical Genetics

Curriculum area	Competence	Exam	Mini-CEX	MSF	CBD	Patient Survey	Generic	PFolio
	at the end of the shift/day with appropriate handover							
	Make adequate arrangements to cover leave.			X			X	
<b>A &amp; B</b>	Recognise the importance of: <ul style="list-style-type: none"> <li>• punctuality</li> <li>• attention to detail.</li> </ul>			X	X		X	
	<b>(ii) Doctor-patient relationship</b>							
<b>Knowledge</b>	Understand all aspects of a professional relationship.			X			X	
	Establish the limiting boundaries surrounding the consultation.		X		X			
	Deal with challenging behaviour in patients which transgress those boundaries, e.g. aggression, violence, racism and sexual harassment.		X	X	X			
<b>Skills</b>	Help the patient appreciate the importance of cooperation between patient and doctor.				X		X	
<b>Skills</b>	Develop a relationship that facilitates solutions to patient's problems.		X	X	X			
	Deal appropriately with behaviour falling outside the boundary of the agreed doctor patient relationship in patients, e.g. aggression, violence, sexual harassment.		X	X	X			
<b>A&amp;B</b>	Adopt a non-discriminatory attitude to all patients and recognise their needs as individuals.		X	X				
	Seek to identify the health care belief of the patient. Acknowledge patient rights to accept or reject advice.		X	X				
	Secure equity of access to health care resources for minority groups.			X	X			X
	Act with compassion at all times			X			X	
	<b>(iii) Recognises own limitations</b>							
<b>Knowledge</b>	Know the extent of one's own limitations and				X		X	

### Assessment Blueprint for Clinical Genetics

Curriculum area	Competence	Exam	Mini-CEX	MSF	CBD	Patient Survey	Generic	PFolio
	know when to ask for advice.							
<b>Skills</b>	Reflection on individual practice				X		X	X
<b>A&amp;B</b>	Be willing to consult and to admit mistakes.			X	X		X	
	<b>(iv) Stress</b>							
<b>Knowledge</b>	Know the effects of stress						X	
	Have knowledge of support facilities for doctors.						X	X
<b>Skills</b>	Develop appropriate coping mechanisms for stress and ability to seek help if appropriate						X	
<b>A&amp;B</b>	Recognise the manifestations of stress on self & others.						X	
	<b>(v) Relevance of outside bodies</b>							
<b>Knowledge</b>	Have an understanding of the relevance to professional life of: The Royal Colleges GMC PMETB Postgraduate Dean Defence unions BMA BSHG / CGS Human Genetic Commission GENCAG Patient representation groups Other organization relevant to genetics	X					X	X
<b>Skills</b>	Recognise situations when appropriate to involve these bodies/individuals.				X		X	
<b>A&amp;B</b>	Be open to constructive criticism			X			X	
	Accept professional regulation.						X	
	Respect the views of patient representation groups.					X		X
	<b>(vi) Personal health</b>							
<b>Knowledge</b>	Know about occupational health services.						X	
	Know about one's responsibilities to the public.						X	

### Assessment Blueprint for Clinical Genetics

Curriculum area	Competence	Exam	Mini-CEX	MSF	CBD	Patient Survey	Generic	PFolio
	Know not to treat oneself or one's family.						X	
<b>Skills</b>	Recognise when personal health takes priority over work pressures and to be able to take the necessary time off.						X	
<b>A&amp;B</b>	Recognise personal health as an important issue.						X	
<b>ETHICS AND LEGAL ISSUES:</b>								
<b>Learning objective:</b> To ensure the trainee has the knowledge and skills to deal appropriately with ethical and legal issues that arise during the management of patients with genetic disorders.								
<b>(i) Informed consent</b>								
<b>Knowledge</b>	Know the process for gaining informed consent	X	X		X		X	
	Understand process of consent for tissue/sample storage and use.	X			X			
	How to gain consent for a research project		X		X		X	X
<b>Skills</b>	Give appropriate information in a manner patients understand and be able to gain informed consent from patients		X	X	X	X		
	Appropriate use of written material				X			
<b>A&amp;B</b>	Consider the patient's needs as an individual				X			
<b>(ii) Confidentiality</b>								
<b>Knowledge</b>	Be aware of relevant strategies to ensure confidentiality.		X		X			
	Be aware of situations when confidentiality might be broken	X			X		X	
<b>Skills</b>	Use and share all information appropriately				X		X	
	Avoid discussing one patient in front of another						X	
	Be prepared to seek patients wishes before disclosing information		X		X		X	
<b>A&amp;B</b>	Respect the right to confidentiality.		X	X	X		X	
<b>(iii) Legal issues relating to Criminal matters</b>								
<b>Knowledge</b>	Know where to seek advice relating to responsibilities in serious criminal matters.				X			

### Assessment Blueprint for Clinical Genetics

Curriculum area	Competence	Exam	Mini-CEX	MSF	CBD	Patient Survey	Generic	PFolio
<b>Skills</b>	Be able to obtain suitable evidence or know whom to consult if in doubt.				X			
<b>A&amp;B</b>	Recognise the importance of legal issues in medical practice and always be ready to seek advice.				X			
	<b>iv) Ethical issues relating particularly to clinical genetics</b>							
<b>Knowledge</b>	Be aware of professional guidelines published by the GMC, BSHG, CGS and other bodies related to clinical genetics	X					X	
<b>Skills</b>	Avoid giving "directive" genetic advice			X	X			
	Be able to communicate ethical issues with patients, colleagues and the public, surrounding: Confidentiality Informed consent Predictive genetic testing Genetic testing of children Population screening for genetic disease Assisted reproduction Prenatal/preimplantation diagnosis Late termination of pregnancy			X	X			X
<b>A &amp; B</b>	Appreciate the diversity of public opinion on ethical and moral aspects of the practice of clinical genetics.			X	X			X
	Respect opinions of patients.			X	X	X		
	Respect the opinion of colleagues.			X	X			
	Be prepared to discuss difficult cases with experienced colleagues and take advice.			X	X			
<b>A&amp;B</b>	Be willing to refer on to a colleague if conflict exists between personal values and those of the patient				X			X
<b>PATIENT EDUCATION AND DISEASE PREVENTION:</b> <b>Learning objective:</b> To ensure that the trainee has the knowledge,								

### Assessment Blueprint for Clinical Genetics

Curriculum area	Competence	Exam	Mini-CEX	MSF	CBD	Patient Survey	Generic	PFolio
	skills and attitudes to be able to educate patients effectively about genetic disease.							
	<ul style="list-style-type: none"> <li>• <b>(i) Educating patients about:</b></li> <li>• <b>disease</b></li> <li>• <b>investigations</b></li> <li>• <b>management</b></li> </ul>							
<b>Knowledge</b>	Know disease course and manifestations.	X	X		X			
	Know investigation procedures including possible alternatives / choices.	X	X		X			
	Be aware of management strategies for genetic disease.	X			X			
<b>Skills</b>	Give information to patients clearly in a manner that they can understand including written information.		X		X	X		
	Encourage questions.		X			X		
	Discuss management plans and follow up arrangements		X		X	X		
<b>A&amp;B</b>	Consider involving patients in developing mutually acceptable investigation plans.				X	X		
	Encourage patients to access: <ul style="list-style-type: none"> <li>• further information</li> <li>• patient support groups</li> </ul>				X	X		
	<b>(ii) Environmental &amp; lifestyle risk factors</b>							
<b>Knowledge</b>	Understand the risk factors that may influence certain genetic diseases, including; Life style, Smoking, Alcohol +Medication	X	X		X			
<b>Skills</b>	Advise on lifestyle changes.		X		X	X		
	Advise on teratogenic potential of medication.		X		X	X		
	Involve other health care workers as appropriate.						X	
<b>A&amp;B</b>	Do not display prejudice		X	X	X		X	

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Curriculum area	Competence	Exam	Mini-CEX	MSF	CBD	Patient Survey	Generic	PFolio
	<b>(iii) Epidemiology &amp; screening</b>							
<b>Knowledge</b>	Know the methods of data collection and their limitations		X		X			X
	Know principles of 1 <sup>o</sup> & 2 <sup>o</sup> prevention & screening.	X			X			
<b>Skills</b>	Assess an individual patient's risk factors.	X	X		X			
	Encourage participation in appropriate disease prevention or screening programmes.		X		X			
<b>A&amp;B</b>	Consider the: <ul style="list-style-type: none"> <li>• positive &amp; negative aspects of prevention</li> <li>• importance of patient confidentiality</li> </ul>		X		X	X	X	
	Respect patient choice.		X		X		X	
<b>WORKING WITH COLLEAGUES:</b> <b>Learning objective:</b> to demonstrate good working relationships with Colleagues.								
<b>Knowledge</b>	Know the roles and responsibilities of team members.			X	X		X	
	Know how a team works effectively.			X	X		X	
	Know the roles of other clinical specialties and their limitations.				X		X	
<b>Knowledge</b>	Know the role of multidisciplinary management in genetic disorders.			X	X		X	
<b>Skills</b>	Show leadership, delegate and supervise safely			X	X		X	
	Be able to communicate effectively.			X			X	
	Handover safely.			X			X	
	Seek advice if unsure.			X	X		X	
	Recognise when input from another specialty is required for individual patients.			X			X	
	Be able to work effectively with GPs, other medical and surgical specialists and other health care professionals			X			X	

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Curriculum area	Competence	Exam	Mini-CEX	MSF	CBD	Patient Survey	Generic	PFolio
<b>A&amp;B</b>	Show respect for others opinions.			X			X	
	Be conscientious and work co-operatively.			X			X	
	Respect colleagues, including non medical professionals, and recognise good advice.			X			X	
	Recognise own limitations.			X			X	
<b>TEAM WORKING &amp; LEADERSHIP SKILLS</b>								
<b>Learning objective:</b> To demonstrate the ability to work in clinical teams and to have the necessary leadership skills.								
<b>Knowledge</b>	Roles & responsibilities of team members.			X			X	
	How a team works.			X			X	
	Ensuring colleagues understand the individual roles and responsibilities of each team member.			X			X	
	Own professional status and specialty						X	
	A knowledge of the field.	X					X	
	The capacity to perceive the need for action and initiate that action			X			X	
<b>Skills</b>	Respect skills and contribution of colleagues			X	X		X	
<b>Skills</b>	To be conscientious and work constructively.			X			X	
	Respect for others opinion.			X			X	
	To recognise your own limitations			X	X		X	X
	Objective setting; Lateral thinking; Planning; Motivating; Organising; Setting example; Negotiation skills.			X			X	
<b>A&amp;B</b>	Recognise own limitations.			X			X	
	Enthusiasm; integrity; courage of convictions; imagination; determination; energy; and professional credibility.			X			X	
<b>TEACHING AND EDUCATIONAL SUPERVISION</b>								
<b>Learning objective:</b> 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)								

### Assessment Blueprint for Clinical Genetics

Curriculum area	Competence	Exam	Mini-CEX	MSF	CBD	Patient Survey	Generic	PFolio
	<b>(i) To have the skills, attitudes and practices of a competent teacher (through participation in a recognized course for medical educators)</b>							
<b>Knowledge</b>	Identify adult learning principles	X						X
	Identify learner needs	X						X
	Identify learning styles.	X						X
	Structure teaching activities for large audiences, small groups and clinic based teaching.							X
	Principles of evaluation.	X						X
<b>Skills</b>	Facilitate learning process.							X
	Identify learning outcomes.							X
	Construct educational objectives.							X
	Design and deliver an effective teaching event.							X
<b>Skills</b>	Communicate effectively with the learners.			X				
	Use effective questioning techniques.		X	X				
	Teach large and small groups effectively.		X	X				
	Select and use appropriate teaching resources.							X
	Give constructive effective feedback.			X				X
	Evaluate programmes and events							X
	Use different media for teaching that are appropriate to the teaching setting.							X
Be able to chair an educational event.			X				X	
<b>A&amp;B</b>	Demonstrate a willingness, enthusiasm and commitment to teach.			X				X
	Show respect for the learner.			X				
	Demonstrate a professional attitude towards teaching.			X				
	Demonstrate a learner centred approach to teaching.			X				
	Seek feedback and demonstrate a willingness to change methods in response to constructive			X				

### Assessment Blueprint for Clinical Genetics

Curriculum area	Competence	Exam	Mini-CEX	MSF	CBD	Patient Survey	Generic	PFolio
	feedback.							
	<b>(ii) Assessment</b>							
<b>Knowledge</b>	Know the principles of assessment	X						X
	Know different assessment methods	X						X
	Define formative and summative assessment	X						X
<b>Skills</b>	Use appropriate assessment methods				X			
	Give constructive, effective feedback			X	X			
<b>A&amp;B</b>	Be honest and objective when assessing performance.			X	X			
	<b>(iii) Appraisal</b>							
<b>Knowledge</b>	Know the principles of appraisal	X						X
	Know the structure of the appraisal interview	X						X
<b>Skills</b>	Participate in effective appraisal		X					X
<b>A&amp;B</b>	Show respect for those participating in appraisal.		X	X				
<b>RESEARCH [Where undertaken]</b>								
<b>Learning objective:</b> 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.								
<b>Knowledge</b>	Be able to set up a hypothesis and test it.							X
	Know how to design a research study.							X
	Know how to use appropriate statistical methods.							X
	Know the principles of research ethics and the role of ethical committees (LREC, MREC).							X
	Know how to write a scientific paper.							X
	How to identify sources of research funding.							X
<b>Skills</b>	Undertake systematic critical review of scientific literature.							X
	Ability to frame questions to be answered by a research project.		X					X

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Curriculum area	Competence	Exam	Mini-CEX	MSF	CBD	Patient Survey	Generic	PFolio
	Develop protocols and methods for research.							X
	Obtain ethical committee approval for a research proposal.							X
	Participate in collaborative research with clinical/scientific colleagues.							X
	Be able to use databases.							X
	Be able to accurately analyse data.	X						
<b>Skills</b>	Write and submit a case report or scientific paper.							X
	Have good written and verbal presentation skills.			X	X			X
<b>A&amp;B</b>	Demonstrate curiosity and a critical spirit of enquiry.				X			X
	Demonstrate the persistence needed to follow a project from inception to completion.							X
	Ensure patient confidentiality.						X	X
	Demonstrate knowledge of the importance of ethical approval and patient consent for clinical research.						X	X
	Humility and the acknowledgement of the contribution of others.			X			X	X
<b>CLINICAL GOVERNANCE</b>								
<b>Learning objective:</b> Demonstrate an understanding of the context, the meaning and the implementation of Clinical Governance.								
	<b>(i) The organisational framework for Clinical Governance at local, health authority and national levels.</b>							
<b>Knowledge</b>	Define the important aspects of Clinical Governance.	X						
	Medical and clinical audit.	X						X
	Research and Development.	X						X
	Integrated care pathways.	X			X			X
	Evidenced based practice.	X			X			
	Clinical effectiveness.	X			X			X

### Assessment Blueprint for Clinical Genetics

Curriculum area	Competence	Exam	Mini-CEX	MSF	CBD	Patient Survey	Generic	PFolio
	Clinical risk systems.	X						X
<b>Knowledge</b>	To define the procedures and the effective action when things go wrong in own practice or that of others.				X			X
	Complaints procedures.				X			X
	Quality assurance schemes in genetic laboratory practice				X			X
<b>Skills</b>	Be an active partaker in clinical governance.							X
	Be able to undertake medical and clinical audit. Be actively involved in audit cycles.							X
	Be active in research and development.							X
	Critically appraise medical data research.				X			X
	Practice evidence based medicine.				X		X	
	Aim for clinical effectiveness (best practice) at all times.				X			X
	Educate self, colleagues and other health care professionals.			X				X
	Be able to handle and deal with complaints in a focused and constructive manner. Learn from complaints			X	X			X
	Develop and institute clinical guidelines and integrated care pathways						X	X
	Be aware of advantages and disadvantages of guidelines.						X	X
	Report and investigate critical incidents.			X				X
Regular review of adverse events and modify practice accordingly.					X	X	X	
<b>Skills</b>	Take appropriate action if you suspect you or a colleague may not be fit to practice.						X	
<b>A&amp;B</b>	Make the care of your patient your first concern.			X			X	
	Respect patient's privacy, dignity and			X			X	

### Assessment Blueprint for Clinical Genetics

Curriculum area	Competence	Exam	Mini-CEX	MSF	CBD	Patient Survey	Generic	PFolio
	confidentiality.							
	Be prepared to learn from mistakes, errors and complaints.			X			X	
	Recognise the importance of team work.			X			X	
	Share best practice with others.			X			X	
	Willingness to cultivate a questioning approach to current practice of clinical genetics and motivation to make improvements.			X	X			X
	<b>(ii) Risk management</b>							
<b>Knowledge</b>	Knowledge of such matters as H&S policy, policies on needle stick injuries, note keeping, communications and staffing numbers.						X	
	Knowledge of risk assessment, perception and relative risk	X			X			X
	Know the complications and side effects of treatments.		X		X			
<b>Skills</b>	Confidently and authoritatively discuss risks with patients and to obtain informed consent		X		X	X		
	Able to balance risks and benefits with patients.		X		X			
<b>A&amp;B</b>	Willingness to respect and accept patients views and choices		X	X	X		X	
	Willingness to be truthful and to admit error to patients, relatives and colleagues			X			X	X
	<b>(iii) Evidence</b>							
<b>Knowledge</b>	Know & understand: the principles of evidence based medicine	X			X		X	
	the types of evidence	X			X			
<b>Skills</b>	Able to critically appraise evidence.				X			X
	Ability to be competent in the use of databases, libraries and the internet.				X		X	X
	Able to discuss the relevance of evidence with		X	X	X			X

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Curriculum area	Competence	Exam	Mini-CEX	MSF	CBD	Patient Survey	Generic	PFolio
	individual patients							
<b>A&amp;B</b>	Display a keenness to use evidence in the support of patient care and own decisions therein.		X		X		X	X
	<b>(iv) Audit</b>							
<b>Knowledge</b>	Know & understand: the audit cycle data sources data confidentiality						X	X
<b>Skills</b>	Involvement in on-going audit.						X	X
	Undertake at least one audit project							X
<b>A&amp;B</b>	Consider the relevance of audit to: benefit patient care clinical governance				X		X	X
	<b>(v) Guidelines</b>							
<b>Knowledge</b>	Know the advantages and disadvantages of guidelines							X
	Methods of determining best practice							X
<b>Skills</b>	Ability to utilise guidelines							X
	Be involved in guideline generation, evaluation and review.							X
<b>A&amp;B</b>	Show regard for individual patient needs when using guidelines		X		X			
	Willingness to use guidelines as appropriate				X			
<b>STRUCTURE OF THE NHS AND THE PRINCIPLES OF MANAGEMENT</b>								
<b>Learning objective:</b> To display knowledge of the structure and organisation of the NHS nationally and locally.								
<b>Knowledge</b>	Know the structure of the NHS, primary care groups, Trusts and Hospital Trusts						X	X
	Know the local Trusts structure including Chief Executives, Medical Directors, Clinical Directors and others.						X	X

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Curriculum area	Competence	Exam	Mini-CEX	MSF	CBD	Patient Survey	Generic	PFolio
	Know the role of postgraduate deaneries, specialist societies, the royal colleges and the general medical council.						X	X
	Know finance issues in general in the Health Service, especially budgetary management.						X	X
	Know the appointments procedures and the importance of equal opportunities.						X	X
	Know of Central Government health regulatory agencies (e.g. NICE, CHI)						X	X
<b>Skills</b>	Develop skills in managing change and managing people.			X				
	Develop leadership skills to play a leading role in developing local genetic services			X				
	Develop interviewing techniques and those required for performance reviews.			X				
	Be able to build a business plan.							X
<b>A&amp;B</b>	Show an awareness of equity in health care access and delivery.						X	X
	Demonstrate an understanding of the importance of a health service for the population.						X	X
	Show respect for others, ensuring equal opportunities.			X			X	X
	Demonstrate a willingness to assume managerial responsibilities.			X				X
<b>INFORMATION TECHNOLOGY, COMPUTER ASSISTED LEARNING AND INFORMATION MANAGEMENT</b> <b>Learning objective:</b> Demonstrate competence in the use and management of health information								
<b>Knowledge</b>	Define how to retrieve and utilize data recorded in clinical systems.				X			X
	Define main local and national projects and initiatives in information technology related to				X			X

### Assessment Blueprint for Clinical Genetics

Curriculum area	Competence	Exam	Mini-CEX	MSF	CBD	Patient Survey	Generic	PFolio
	genetics							
	Demonstrate an understanding of the range of possible uses for clinical data and information and appreciate the dangers and benefits of aggregating clinical data.				X		X	X
	To understand implications of the Data Protection Act for patient confidentiality, including genetic registers.				X		X	
<b>Skills</b>	Demonstrate competent use of database, word processing and statistics programmes.				X		X	X
	Undertake effective literature searches.						X	
	Access genetic web sites and specialist databases to undertake searches.		X		X			
<b>Skills</b>	Use commercial software packages, including CYRILLIC.				X			X
	To appraise available software.				X			X
	To apply the principles of confidentiality and their implementation in terms of clinical practice in the context of information technology.						X	X
	Produce effective computer assisted presentations.				X			X
<b>A&amp;B</b>	Demonstrate the acquisition of new attitudes in patient consultations in order to make maximum use of information technology.		X		X			X
	Be willing to offer advice to lay person on access to appropriate Internet sources and support groups.		X		X			X
	Adopt proactive and enquiring attitude to new technology.				X			X