

Curriculum for Clinical Genetics Training Implementation August 2021

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1. Introduction

The speciality of Clinical Genetics is evolving at a rapid pace, driven in part by enormous advances in the technology available to make genetic or genomic diagnoses. This in turn has meant that testing has shifted from a focused enquiry of single genes to genome wide analyses with subsequent data interrogation.

Clinical Genetics is the discipline that is concerned with the diagnosis and management of genetic and genomic disorders and birth defects and with counselling of family members regarding risk, surveillance or prevention. Physicians in this speciality work in multidisciplinary regional centres in close collaboration with genetic counsellors, laboratory and academic colleagues.

The purpose of the Clinical Genetics curriculum is to produce doctors with the generic professional and speciality specific capabilities needed to undertake a consultant role in a Regional Clinical Genetics Service. Clinical Genetics trainees will be entrusted to practice within a Regional Clinical Genetics Service operating both within inpatient and outpatient settings and also in a liaison capacity with genomic laboratory services, which may in some instances be local and in others may be regional or national. Training in Clinical Genetics will normally take place over an indicative period of 4 years and will include mandatory training in all core areas of practice. It will also include training in genomic variant interpretation and bioinformatics as applied to the clinical setting and will produce doctors able to apply for consultant roles in the discipline.

The curriculum for Clinical Genetics has been developed with input from trainees, consultants actively involved in delivering teaching and training across the UK, service representatives and lay persons. This has been in liaison with the JRCPTB, Clinical Genetics Specialist Advisory Committee, British Society for Genomic Medicine (BSGM) and The Joint Committee of Genomics in Medicine.

The Shape of Training (SoT) review was a catalyst for reform of postgraduate training of all doctors to ensure it is more patient focused, more general (especially in the early years) and has more flexibility of career structure. For physician training, the views and recommendations of SoT were similar to those of the Future Hospital Commission and the Francis report.

A further driver for change was the GMC review of the curricula and assessment standards and introduction of the GPC framework. From May 2017, all postgraduate curricula should be based on higher level learning outcomes and must incorporate the generic professional capabilities. A fundamental component of the GPCs is ensuring that the patient is at the centre of any consultation and decision making.

A training in Clinical Genetics is a training that includes (but is not limited to) the diagnosis and management of rare disease. Although rare diseases are, by definition, not present

individually in many in the population, if taken as a group together rare diseases affect one person in seventeen [1]. With the enormous changes in genomic technologies in the last few years, the biological pathways underlying these disorders are slowly being elucidated and this will lead to more effective management strategies and more individualised patient care pathways. Whilst in general the burden of genetics disease is distributed throughout populations, there are some specific communities within the four countries to which this document applies, who have an increased burden of some specific disorders, or classes of disorder, and this curriculum addresses these needs.

‘The Evolving Role Of The Clinical Geneticist’ [2] addresses some of the changes that a clinical geneticist is facing, including the need for skills in genomic variant interpretation and bioinformatics and thus a need for increased utilisation of up to date web-based interfaces and IT applications.

As the speed of genomic sequencing increases and the cost reduces, many more patients will have testing relevant to their care and geneticists will need to interact with more specialities across the healthcare sector. Increasingly patients will have broad genomic analysis undertaken as part of their care and the clinical geneticist will assist other generalist and specialist doctors in assessing the clinical relevance of the many genomic variants that result from these new approaches. Whilst mainstreaming of some genomic testing will be possible, clinical geneticists will be crucial to ensuring the correct interpretation of the results of broad genomic testing. Trainees in the discipline will have extensive training in this key role, which will form an increasing part of the workload of a clinical geneticist.

As genomic sequencing and interpretation becomes more streamlined, its general applicability to some acute care will increase such that some key management decisions in acute settings will involve the clinical geneticist. In addition, genomic susceptibilities in chronic disease will mean that clinical geneticists will play a role in community healthcare settings, providing individualised patient management advice. Whilst geneticists will mainly be involved in an advisory role in the provision of acute hospital care and general medical services, their ongoing role will support the principles set out in the Future Hospital Commission.

Clinical Geneticists will still retain a primary diagnostic role but are likely to be more integrated into the discipline where they have particular expertise. There will be increased involvement in MDT clinics and meetings for patient care, particularly for individuals with complex multisystem disorders.

2. Purpose

2.1 Purpose of the curriculum

Many of the concepts and ways of working will be new to the trainee whichever stem has begun his or her training, and many of the clinical genetics speciality capabilities may not be fully reached until the later stages of training. Completion of early training in many different disciplines (including IM stage 1 and paediatrics stage 1) will be the first stage of training and a critical progression point. Trainees will need to have gained a wide range of clinical

skills to deal with the broad age range seen in genetics consultations and the diverse and rare presentations encountered. This variation could be from a family where adult onset cancers have been present, to a couple dealing with a severely disabled child, to a pregnant mother with abnormal scan findings. Trainees will also need to have gained the key patient facing skills required to hold a full consultation and be able to synthesise history and clinical findings to direct onward management in this broad variety of clinical situations. Therefore, trainees will need to have gained key capabilities in clinical prioritisation, holistic patient care, continuity of care, outpatient care, management of long term conditions and experience of MDT working from their early training. Entry into clinical genetics higher specialist training will require completion of one of the following programmes: Internal Medicine Training – stage one (2-years), ACCS- Internal Medicine (3-years), Level 1 Paediatrics training (3 years), Core Surgical Training (2 years), Two years), Core Psychiatry Training (3 years), Basic (ST1 and ST2) Obstetrics and Gynaecology training (2 years), ST1 and ST2 of Ophthalmic Specialist Training, or completion of a General Practice specialty training programme (3 years) which includes completion of the associated high stakes assessments which will provide the skills described above and are detailed in the training pathway diagram below. Progression through clinical genetics training will require gaining all 6 specialist capabilities which build upon those already achieved in foundation training and early broad-based speciality training. Clinical genetics trainees will not be required to provide either medical or paediatric acute unselected care. It is anticipated that most trainees will be entrusted to manage general and cancer genomics consultations by the end of the second year of clinical genetics training.

In order to gain appropriate skills in genomic interpretation and achieve the clinical genetics capabilities trainees may wish to complete a post graduate certificate in genomics during their training by undertaking appropriate modules of an approved genomics MSc course. Some trainees may however prefer to gain these skills, using online resources, by attending alternative courses with major emphasis on genomic variant interpretation or via appropriate laboratory attachments.

Scope of practice

The capabilities to be achieved in this curriculum build on those achieved during early broad-based speciality training, such as internal medicine stage 1. The scope of clinical genetics practice requires a high degree of diagnostic ability underpinned by excellence in the basic sciences pertinent to the discipline, and a willingness to be involved in translational research. Clinical genetics is changing very rapidly and trainees must be able to adapt to take into account new technologies that will alter clinical practice. Trainees will also need to have a wide variety of clinical skills that will allow them to work with patients and families from all age ranges and communicate effectively in a diversity of life-altering situations from a critical decision point within a pregnancy to a family struggling to understand a hereditary form of cancer. Doctors in clinical genetics have always worked very closely with genetic counsellors, who will continue to work with families when a diagnosis of a genetic disease has been made and cascade testing or further counselling of family members is required. The clinical geneticist also learns to work closely with laboratory colleagues to ensure an optimum interpretation of genetic and genomic variants in a given clinical situation. In addition, a clinical genetics trainee will learn effective interaction with research teams for optimal patient benefit and become involved in

recruitment to studies and therapeutic trials. As genomics increasingly underpins many other areas of medical practice, physicians involved in the discipline will have an important educational role both in clinical practice, but also in public education and public debate surrounding ethical issues arising from new developments.

2.2 High level learning outcomes – capabilities in practice (CiPs)

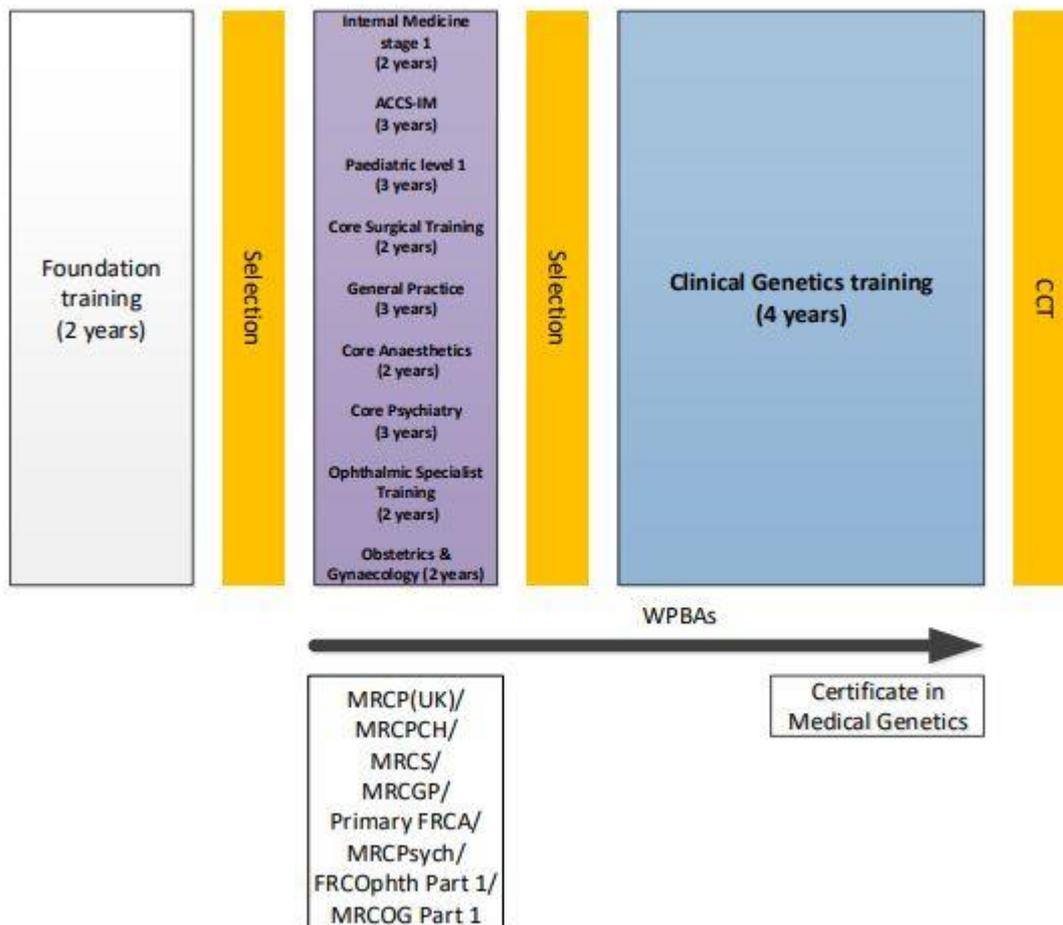
The 6 CiPs for clinical genetics describe the scope of professional tasks to be entrusted to that doctor by the end of training. Each CiP has a set of descriptors associated with that activity or task. The descriptors are intended to help trainers and trainees recognise the minimum level of knowledge, skill and attitude that should be demonstrated for an entrustment decision to be made. By completion of clinical genetics training the doctor must have demonstrated that they are capable of unsupervised practice in all 6 speciality CiPs.

The clinical genetics CiPs describe the clinical tasks or activities which are essential to the practice of the specialty. They have been mapped to the GPC domains and subsections to reflect the professional generic capabilities required to undertake the clinical tasks. Satisfactory sign off requires demonstration that, for each of the CiPs, the doctor in training's performance meets or exceeds the minimum expected level for completion of training, as defined in the curriculum.

Learning outcomes – capabilities in practice (CiPs)
Generic CiPs
<ol style="list-style-type: none"> 1. Able to successfully function within NHS organisational and management systems 2. Able to deal with ethical and legal issues related to clinical practice 3. Communicates effectively and is able to share decision making, while maintaining appropriate situational awareness, professional behaviour and professional judgement 4. Is focused on patient safety and delivers effective quality improvement in patient care 5. Carrying out research and managing data appropriately 6. Acting as a clinical teacher and clinical supervisor
Specialty CiPs
<ol style="list-style-type: none"> 1. Managing a comprehensive genetic medicine service for both inpatients and outpatients 2. Working within multidisciplinary teams and consultations related to the management and treatment of complex genetic disorders. 3. Managing predictive genetic testing and advising on cascade genetic testing in families. 4. Managing storage and testing of genetic material in the prenatal and post mortem settings.

5. Interrogating and interpreting genetic data and communicating effectively with laboratory colleagues.
6. Contributing to genetic research and clinical trials.

2.3 Training pathway



2.4 Duration of training

Training in Clinical Genetics will normally take place over an indicative period of 4 years and will include mandatory training in all core areas of practice. It will also include training in genomic variant interpretation and bioinformatics as applied to the clinical setting and will produce doctors able to apply for consultant roles in the discipline. The Shape of Training review has provided a further catalyst for change in all postgraduate medical education and will ensure that care is more patient focused. The clinical geneticist will be key in many areas of medicine in helping to apply the increased patient specific knowledge that will result from broad based genomic testing. Another driver from The Shape of Training review is to provide greater flexibility of career structure for doctors and with an emphasis on generic capabilities across many disciplines in early medical training. With this in place, it will be possible to enter a clinical genetics training from a greater variety of early broad-based speciality training including medical, paediatric, surgical, psychiatric, obstetrics and gynaecology, ophthalmology and general practice bases. Entry

into clinical genetics specialist training will require full MRCP(UK) or MRCPCH, MRCS, MRCPGP, primary FRCA, MRCPsych, part 1 MRCOG, part 1 FRCOphth.

There will be options for those trainees who demonstrate exceptionally rapid development and acquisition of capabilities to complete training more rapidly than the current indicative time although it is recognised that clinical experience is a fundamental aspect of development as a good physician (guidance on completing training early will be available on the [JRCPTB website](#)). There may also be a small number of trainees who develop more slowly and will require an extension of training in line the Reference Guide for Postgraduate Specialty Training in the UK (The Gold Guide)¹.

2.5 Flexibility and accreditation of transferable capabilities

The curriculum incorporates and emphasises the importance of the generic professional capabilities (GPCs). GPCs will promote flexibility in postgraduate training as these common capabilities can be transferred from specialty to specialty. In addition, the curriculum supports flexibility for trainees to move between these specialties without needing to repeat aspects of training. The curriculum supports the accreditation of transferrable competencies (using the Academy framework).

Recognition of training from some other higher specialist training programmes to clinical genetics will be possible. Competencies may count towards a CCT in Clinical Genetics if gained from specialist training in general internal medicine stage 2, any higher specialist medicine programme (JRCPTB specialty), higher specialty training in paediatrics or ophthalmology, provided appropriate capabilities have been reached. These will be assessed on a case by case basis. As an example, a trainee who has undertaken 6 months or greater training in medical oncology may have relevant experience recognised if they choose to transfer (via competitive entry) into a Clinical Genetics training, and may reduce their indicative training time to 3.5 years.

2.6 Less than full time training

Trainees are entitled to opt for less than full time training programmes. Less than full time 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.

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 in accordance with the Gold Guide.

This purpose statement has been endorsed by the GMC's Curriculum Oversight Group and confirmed as meeting the needs of the health services of the countries of the UK.

¹ [A Reference Guide for Postgraduate Specialty Training in the UK](#)

2.7 Generic Professional Capabilities and Good Medical Practice

The GMC has developed the Generic professional capabilities (GPC) framework² with the Academy of Medical Royal Colleges (AoMRC) to describe the fundamental, career-long, generic capabilities required of every doctor. The framework describes the requirement to develop and maintain key professional values and behaviours, knowledge, and skills, using a common language. GPCs also represent a system-wide, regulatory response to the most common contemporary concerns about patient safety and fitness to practise within the medical profession. The framework will be relevant at all stages of medical education, training and practice.

The nine domains of the GMC's Generic Professional Capabilities



Good medical practice (GMP)³ is embedded at the heart of the GPC framework. In describing the principles, duties and responsibilities of doctors the GPC framework articulates GMP as a series of achievable educational outcomes to enable curriculum design and assessment.

The GPC framework describes nine domains with associated descriptor outlining the 'minimum common regulatory requirement' of performance and professional behaviour for those completing a CCT or its equivalent. These attributes are common, minimum and generic standards expected of all medical practitioners achieving a CCT or its equivalent.

² [Generic professional capabilities framework](#)

³ [Good Medical Practice](#)

The nine domains and subsections of the GPC framework are directly identifiable in the curriculum. They are mapped to each of the generic and specialty CiPs, which are in turn mapped to the assessment blueprints. This is to emphasise those core professional capabilities that are essential to safe clinical practice and that they must be demonstrated at every stage of training as part of the holistic development of responsible professionals.

This approach will allow early detection of issues most likely to be associated with fitness to practise and to minimise the possibility that any deficit is identified during the final phases of training.

3. Content of Learning

The clinical genetics curriculum provides a broad based training in all areas of relevant clinical practice, across a wide age range, and in the interpretation of genomic variants pertinent to medical care. In order to practice as a clinical geneticist, an individual must gain appropriate experience in the clinical areas that are core to the discipline. This must include training in bioinformatics, omics and variant interpretation as applied to the clinical setting and in addition the genomics of common and rare disease, the genomics of developmental disorders, cancer genetics, cardiac genetics, neurogenetics and prenatal genetics. As a practicing geneticist a physician would often be a key clinician involved in such clinical episodes. However there are many other areas of clinical practice where clinical geneticists have roles to play in care, but would be less likely to be the lead clinician involved in the episode. For these areas of practice some experience should be gained during clinical training, but a detailed knowledge of the genomics relevant to clinical care in these areas could be obtained post CCT where relevant for individual practice.

Clinical geneticists have always been involved in management of lifetime risk and chronic disorders and provided care through the full spectrum of ages. This curriculum will ensure that trainees for the future are fully trained in the long-term management of genomic disorders, including documenting the natural history, managing its medical complications and advising at risk relatives.

The curriculum is spiral, and topics and themes will be revisited to expand understanding and expertise. The level of entrustment for capabilities in practice (CiPs) will increase as an individual progresses from needing direct supervision to able to be entrusted to act unsupervised.

3.1 Capabilities in practice (CiPs)

CiPs describe the professional tasks or work within the scope of the specialty. CiPs are based on the concept of entrustable professional activities⁴ which use the professional judgement of appropriately trained, expert assessors as a defensible way of forming global judgements of professional performance.

⁴ [Nuts and bolts of entrustable professional activities](#)

Each CiP has a set of descriptors associated with that activity or task. Descriptors are intended to help trainees and trainers recognise the knowledge, skills and attitudes which should be demonstrated. Doctors in training may use these capabilities to provide evidence of how their performance meets or exceeds the minimum expected level of performance for their year of training. The descriptors are not a comprehensive list and there are many more examples that would provide equally valid evidence of performance.

Many of the CiP descriptors refer to patient centred care and shared decision making. This is to emphasise the importance of patients being at the centre of decisions about their own treatment and care, by exploring care or treatment options and their risks and benefits and discussing choices available.

Additionally, the CiPs repeatedly refer to the need to demonstrate professional behaviour with regard to patients, carers, colleagues and others. Good doctors work in partnership with patients and respect their rights to privacy and dignity. They treat each patient as an individual. They do their best to make sure all patients receive good care and treatment that will support them to live as well as possible, whatever their illness or disability. Appropriate professional behaviour should reflect the principles of GMP and the GPC framework.

In order to complete training and be recommended to the GMC for the award of CCT and entry to the specialist register, the doctor must demonstrate that they are capable of unsupervised practice in all generic and specialty CiPs. Once a trainee has achieved level 4 sign off for a CiP it will not be necessary to repeat assessment of that CiP if capability is maintained (in line with standard professional conduct).

This section of the curriculum details the six generic CiPs and six of specialty CiPs for Clinical Genetics. The expected levels of performance, mapping to relevant GPCs and the evidence that may be used to make an entrustment decision are given for each CiP.

The list of evidence for each CiP is not prescriptive and other types of evidence may be equally valid for that CiP.

3.2 Generic capabilities in practice

The six generic CiPs cover the universal requirements of all specialties as described in GMP and the GPC framework. Assessment of the generic CiPs will be underpinned by the descriptors for the nine GPC domains and evidenced against the performance and behaviour expected at that stage of training. Satisfactory sign off will indicate that there are no concerns. It will not be necessary to assign a level of supervision for these non-clinical CiPs.

In order to ensure consistency and transferability, the generic CiPs have been grouped under the GMP-aligned categories used in the Foundation Programme curriculum plus an additional category for wider professional practice:

- professional behaviour and trust
- communication, team-working and leadership
- safety and quality

- wider professional practice.

For each generic CiP there is a set of descriptors of the observable skills and behaviours which would demonstrate that a trainee has met the minimum level expected. The descriptors are not a comprehensive list and there may be more examples that would provide equally valid evidence of performance.

KEY

CbD	Case-based discussion	DOPS	Direct observation of procedural skills
GCP	Good Clinical Practice	MCR	Multiple consultant report
Mini-CEX	Mini-clinical evaluation exercise	PS	Patient survey
MSF	Multi source feedback	TO	Teaching observation
QIPAT	Quality improvement project assessment tool		

Generic capabilities in practice (CiPs)

Category 1: Professional behaviour and trust

1. Able to function successfully within NHS organisational and management systems

Descriptors	<ul style="list-style-type: none"> • Aware of and adheres to the GMC professional requirements • Aware of public health issues including population health, social detriments of health and global health perspectives • Demonstrates effective clinical leadership • Demonstrates promotion of an open and transparent culture • Keeps practice up to date through learning and teaching • Demonstrates engagement in career planning • Demonstrates capabilities in dealing with complexity and uncertainty • Aware of the role of and processes for commissioning • Aware of the need to use resources wisely
GPCs	<p>Domain 1: Professional values and behaviours</p> <p>Domain 3: Professional knowledge</p> <ul style="list-style-type: none"> • professional requirements • national legislative requirements • the health service and healthcare systems in the four countries <p>Domain 9: Capabilities in research and scholarship</p>
Evidence to inform decision	<p>MCR</p> <p>MSF</p> <p>Active role in governance structures</p> <p>Management course</p> <p>End of placement reports</p>

2. Able to deal with ethical and legal issues related to clinical practice

Descriptors	<ul style="list-style-type: none"> • Aware of national legislation and legal responsibilities, including safeguarding vulnerable groups • Behaves in accordance with ethical and legal requirements • Demonstrates ability to offer apology or explanation when appropriate • Demonstrates ability to lead the clinical team in ensuring that medical legal factors are considered openly and consistently
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GPCs	<p>Domain 3: Professional knowledge</p> <ul style="list-style-type: none"> • professional requirements • national legislative requirements • the health service and healthcare systems in the four countries <p>Domain 4: Capabilities in health promotion and illness prevention</p> <p>Domain 7: Capabilities in safeguarding vulnerable groups</p> <p>Domain 8: Capabilities in education and training</p> <p>Domain 9: Capabilities in research and scholarship</p>
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Evidence to inform decision	<p>MCR</p> <p>MSF</p> <p>CbD</p> <p>DOPS</p> <p>Mini-CEX</p> <p>ALS certificate</p> <p>End of life care and capacity assessment</p> <p>End of placement reports</p>
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Category 2: Communication, teamworking and leadership

3. Communicates effectively and is able to share decision making, while maintaining appropriate situational awareness, professional behaviour and professional judgement

Descriptors	<ul style="list-style-type: none"> • Communicates clearly with patients and carers in a variety of settings • Communicates effectively with clinical and other professional colleagues • Identifies and manages barriers to communication (eg cognitive impairment, speech and hearing problems, capacity issues) • Demonstrates effective consultation skills including effective verbal and nonverbal interpersonal skills • Shares decision making by informing the patient, prioritising the patient’s wishes, and respecting the patient’s beliefs, concerns and expectations • Shares decision making with children and young people • Applies management and team working skills appropriately, including influencing, negotiating, re-assessing priorities and effectively managing complex, dynamic situations
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GPCs	Domain 2: Professional skills <ul style="list-style-type: none"> • practical skills • communication and interpersonal skills • dealing with complexity and uncertainty • clinical skills (<i>history taking, diagnosis and medical management; consent; humane interventions; prescribing medicines safely; using medical devices safely; infection control and communicable disease</i>) Domain 5: Capabilities in leadership and teamworking
Evidence to inform decision	MCR MSF PS End of placement reports ES report
Category 3: Safety and quality	
4. Is focused on patient safety and delivers effective quality improvement in patient care	
Descriptors	<ul style="list-style-type: none"> • Makes patient safety a priority in clinical practice • Raises and escalates concerns where there is an issue with patient safety or quality of care • Demonstrates commitment to learning from patient safety investigations and complaints • Shares good practice appropriately • Contributes to and delivers quality improvement • Understands basic Human Factors principles and practice at individual, team, organisational and system levels • Understands the importance of non-technical skills and crisis resource management • Recognises and works within limit of personal competence • Avoids organising unnecessary investigations or prescribing poorly evidenced treatments
GPCs	Domain 1: Professional values and behaviours Domain 2: Professional skills <ul style="list-style-type: none"> • practical skills • communication and interpersonal skills • dealing with complexity and uncertainty • clinical skills (<i>history taking, diagnosis and medical management; consent; humane interventions; prescribing medicines safely; using medical devices safely; infection control and communicable disease</i>) Domain 3: Professional knowledge <ul style="list-style-type: none"> • professional requirements • national legislative requirements • the health service and healthcare systems in the four countries Domain 4: Capabilities in health promotion and illness prevention Domain 5: Capabilities in leadership and teamworking Domain 6: Capabilities in patient safety and quality improvement <ul style="list-style-type: none"> • patient safety

	<ul style="list-style-type: none">• quality improvement
Evidence to inform decision	MCR MSF QIPAT End of placement reports

Category 4: Wider professional practice

5. Carrying out research and managing data appropriately

Descriptors	<ul style="list-style-type: none"> • Manages clinical information/data appropriately • Understands principles of research and academic writing • Demonstrates ability to carry out critical appraisal of the literature • Understands the role of evidence in clinical practice and demonstrates shared decision making with patients • Demonstrates appropriate knowledge of research methods, including qualitative and quantitative approaches in scientific enquiry • Demonstrates appropriate knowledge of research principles and concepts and the translation of research into practice • Follows guidelines on ethical conduct in research and consent for research • Understands public health epidemiology and global health patterns • Recognises potential of applied informatics, genomics, stratified risk and personalised medicine and seeks advice for patient benefit when appropriate
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GPCs	<p>Domain 3: Professional knowledge</p> <ul style="list-style-type: none"> • professional requirements • national legislative requirements • the health service and healthcare systems in the four countries <p>Domain 7: Capabilities in safeguarding vulnerable groups</p> <p>Domain 9: Capabilities in research and scholarship</p>
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Evidence to inform decision	<p>MCR</p> <p>MSF</p> <p>GCP certificate (if involved in clinical research)</p> <p>Evidence of literature search and critical appraisal of research</p> <p>Use of clinical guidelines</p> <p>Quality improvement and audit</p> <p>Evidence of research activity</p> <p>End of placement reports</p>
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6. Acting as a clinical teacher and clinical supervisor

Descriptors	<ul style="list-style-type: none"> • Delivers effective teaching and training to medical students, junior doctors and other health care professionals • Delivers effective feedback with action plan • Able to supervise less experienced trainees in their clinical assessment and management of patients • Able to supervise less experienced trainees in carrying out appropriate practical procedures <p><input type="checkbox"/> Able to act a clinical supervisor to doctors in earlier stages of training</p>
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GPCs	Domain 1: Professional values and behaviours Domain 8: Capabilities in education and training
Evidence to inform decision	MCR MSF TO Relevant training course End of placement reports

3.3 Specialty capabilities in practice

The specialty CiPs describe the clinical tasks or activities which are essential to the practice of Clinical Genetics. The CiPs have been mapped to the nine GPC domains to reflect the professional generic capabilities required to undertake the clinical tasks.

Satisfactory sign off will require educational supervisors to make entrustment decisions on the level of supervision required for each CiP and if this is satisfactory for the stage of training, the trainee can progress. More detail is provided in the programme of assessment section of the curriculum.

KEY

KBA Clinical Genetics	Certificate Examination in Medical Genetics	DOPS	Direct observation of procedural skills
CbD	Case-based discussion	PG Cert	Post graduate Certificate in Genomics
GCP	Good Clinical Practice	MCR	Multiple consultant report
Mini-CEX	Mini-clinical evaluation exercise	PS	Patient survey
MSF	Multi source feedback	TO	Teaching observation
QIPAT	Quality improvement project assessment tool		

Specialty CiPs

1. Managing a comprehensive genetic medicine service for both inpatients and outpatients

Descriptors	<ul style="list-style-type: none"> • Conducts essential pre-clinic preparation • Constructs an accurate genogram • Assesses patient and family history • Undertakes clinical examination relevant to clinical condition • Discusses utility of genetic testing including limitations/uncertainties • Takes appropriate consent for testing/storage of genetic material • Manages confidentiality relating to genetic information/testing in family • Diagnoses genetic disorders based on clinical findings and/or genomic results • Discusses genetic test results and recommends/institutes appropriate clinical management for immediate and longitudinal care of family members. • Documents accurately the outcome and implications of the genetic episode
GPCs	<p>Domain 1: Professional values and Behaviours</p> <p>Domain 2: Professional Skills</p> <p style="padding-left: 40px;">Practical skills</p> <p style="padding-left: 40px;">Communication and interpersonal skills</p> <p style="padding-left: 40px;">Dealing with complexity and uncertainty</p> <p style="padding-left: 40px;">Clinical Skills</p> <p>Domain 7: Capabilities in safeguarding vulnerable groups</p>
Evidence to inform decision	<p>Mini-CEX</p> <p>MSF</p> <p>CbD</p> <p>MCR</p> <p>PS</p> <p>End of placement reports</p> <p>ES reports</p> <p>Genomics PG Cert or MSc modules</p> <p>KBA Clinical Genetics</p>
2. Working within multidisciplinary teams and consultations related to the management and treatment of complex genetic disorders.	
Descriptors	<ul style="list-style-type: none"> • Adapts the genetic consultation to participate effectively in the multidisciplinary clinic setting • Recognises and effectively communicates the value of the genetic contribution to colleagues and patients • Manages genetic investigations including the communication of patient test results

	<ul style="list-style-type: none"> • Demonstrates appropriate knowledge of the specific speciality with the MDT and makes relevant and focused contribution • Integrates genetic investigations, results and counselling into the patient pathway effectively
GPCs	Domain 1: Professional values and behaviours Domain 2: Professional Skills Domain 5: Capabilities in leadership and team working
Evidence to inform decision	MSF Mini-CEX Cbd End of placement reports Speciality training courses Use of clinical guidelines Genomics PG Cert or MSc modules KBA Clinical Genetics
3. Managing predictive genetic testing and advising on cascade genetic testing in families.	
Descriptors	<ul style="list-style-type: none"> • Discriminate between diagnostic and predictive genetic testing pathways • Evaluates and communicates the advantages and disadvantages of a predictive genetic test • Demonstrates understanding of lasting impact of predictive genetic test results • Demonstrates ability to accurately calculate or otherwise assess the risks of a genetic disorder to a family member • Implements an appropriate management plan to allow testing/screening for family members • Demonstrates effective working with speciality and genetic counselling colleagues to plan cascade testing or screening
GPCs	Domain 1: Professional values and behaviours Domain 2: Professional skills Dealing with complexity and uncertainty History taking, diagnosis, medical management and consent Domain 4: Capabilities in health promotion and illness prevention Domain 6: Capabilities in patient safety and quality improvement
Evidence to inform decision	Mini CEX CBD PS MCR Speciality training courses

	<p>Counselling module of Genomics PGCert End of placement report ES reports</p>
<p>4. Managing storage and testing of genetic material in the prenatal and post mortem settings.</p>	
<p>Descriptors</p>	<ul style="list-style-type: none"> • Demonstrates ability to apply genetic principles in the prenatal setting and for pre-implantation genetic diagnosis and non-invasive prenatal diagnosis when applicable • Manages the appropriate handling of genetic data/material after death of patient with a genetic condition • Demonstrates accurate understanding of the legal framework relating to storage and testing of genetic material including implications for families
<p>GPCs</p>	<p>Domain 1: Professional Values and behaviours Domain 2: Professional skills Domain 3: Professional knowledge Domain 6: Capabilities in patient safety and quality improvement.</p>
<p>Evidence to inform decision</p>	<p>CbD Speciality training courses MSF MCR End of placement reports ES reports</p>
<p>5. Interrogating and interpreting genetic data and communicating effectively with laboratory colleagues.</p>	
<p>Descriptors</p>	<ul style="list-style-type: none"> • Demonstrates understanding of genetic architecture and the cellular and molecular mechanisms that underpin inheritance in man • Demonstrates understanding and application of the laboratory techniques that underpin current genetic testing • Demonstrates ability to interrogate bioinformatics databases to aid in the interpretation of genomic variants • Discusses the genetic testing strategy with clinical and scientific colleagues • Demonstrates the ability to critically appraise cytogenetic and molecular reports and assess their relevance for patient care
<p>GPCs</p>	<p>Domain 1: Professional Values and behaviours Domain 2: Professional skills Communication and interpersonal skills Dealing with complexity and uncertainty Domain 5: Capabilities in leadership and team working</p>

	Domain 6: Capabilities in patient safety and quality improvement Domain 9: Capabilities in research and scholarship
Evidence to inform decision	MSF CbD GCP Genomics PG Cert KBA Clinical Genetics Speciality training courses Evidence of research activity
6. Contributing to Genetic Research and clinical trials	
Descriptors	<ul style="list-style-type: none"> • Recognises unsolved genetic questions at the limit of knowledge • Contributes to and collaborates in research studies for patient benefit • Demonstrates understanding of genome-based therapies • Seeks opportunities to develop new strategies that will benefit patients in the future
GPCs	Domain 4: Capabilities in health promotion and illness prevention Domain 9: Capabilities in research and scholarship
Evidence to inform decision	CbD GCP Genomics PG Cert KBA Clinical Genetics Evidence of research activity MD or PhD

3.4 Presentations and conditions

The table below details the key presentations and conditions of Clinical Genetics. Each of these should be regarded as a clinical context in which trainees should be able to demonstrate CiPs and GPCs. In this spiral curriculum, trainees will expand and develop the knowledge, skills and attitudes around managing patients with these conditions and presentations. The patient should always be at the centre of knowledge, learning and care.

Trainees must demonstrate core bedside skills, including information gathering through history and physical examination and information sharing with patients, families and colleagues.

Treatment care and strategy covers how a doctor selects interventions or screening strategies for a patient. It also covers broader aspects of care, including involvement of other professionals or services.

Particular presentations, conditions and issues are listed either because they are common or serious (having high morbidity, mortality and/or serious implications for treatment or public health).

For each condition/presentation, trainees will need to be familiar with such aspects as aetiology, epidemiology, clinical features, investigation, management and prognosis. Our approach is to provide general guidance and not exhaustive detail, which would inevitably become out of date.

System/Specialty F	Presentations	Conditions/Issues
General genetics	<ul style="list-style-type: none"> • Developmental delay and intellectual disability • Significant behavioural difficulties including autism • Visual impairment • Hearing loss • Consanguinity • Common autosomal recessive disorders • Common autosomal dominant disorders 	<ul style="list-style-type: none"> • Disorders caused by copy number variation including deletion and microdeletion syndromes and duplication and microduplication syndromes. • Chromosome translocations • Polycystic kidney disease • Retinitis Pigmentosa • Hyperlipidaemia • Diabetes mellitus • Cystic fibrosis • Alpha 1 antitrypsin deficiency • Haemochromatosis • Hereditary haemorrhagic telangiectasia • Haemoglobinopathies • Haemophilia • Stickler syndrome
Cancer genetics	<ul style="list-style-type: none"> • Family history of common cancers e.g. breast, ovarian and colorectal • Personal and/or family history of rare cancers • Family history of known familial cancer syndromes 	<ul style="list-style-type: none"> • BRCA1 • BRCA2 • Lynch syndrome • PTEN • Familial Polyposis Syndromes • Gorlin syndrome • Li Fraumeni syndrome

	<ul style="list-style-type: none"> • Personal history of cancer with abnormal somatic or germline test results 	<ul style="list-style-type: none"> • Multiple endocrine neoplasia • Neurofibromatosis type 2 • Von Hippel Lindau • Pheochromocytoma • Wilms tumour
Cardiac genetics	<ul style="list-style-type: none"> • Features suggestive of a connective tissue disorder • Aortic dissection, dilatation or rupture • Sudden cardiac death • Family history of congenital heart disease • Family history of cardiomyopathy (hypertrophic, dilated or arrhythmogenic) • Familial arrhythmias • Infantile cardiomyopathy (hypertrophic, dilated, non-compaction) • Cardiomyopathy in context of likely mitochondrial or metabolic disease 	<ul style="list-style-type: none"> • Marfan syndrome • Loeys Dietz syndromes • Non-syndromic familial aortopathy • Ehlers Danlos syndromes • Familial Dilated Cardiomyopathy • LMNA • Hypertrophic Cardiomyopathy • Non-compaction Cardiomyopathy • Arrhythmogenic Cardiomyopathy • Metabolic Cardiomyopathy • Mitochondrial Cardiomyopathy • Long QT syndrome • Brugada syndrome • CPVT
Neurogenetics	<ul style="list-style-type: none"> • Infantile hypotonia • Epileptic encephalopathy • Structural brain anomalies • Neuronal migration disorders • Leukodystrophy • Seizure disorders • Microcephaly and macrocephaly 	<ul style="list-style-type: none"> • Prader willi syndrome • Angleman syndrome • Spinal muscular atrophy • Myotonic Dystrophy • HSMN type 1a • Huntington Disease • Duchenne and Becker muscular dystrophy • Facioscapulohumeral muscular dystrophy • Rett syndrome

	<ul style="list-style-type: none"> • Developmental regression • Movement disorders • Myopathies and muscular dystrophies • Hereditary neuropathies and SMA's • Ataxias • Hereditary Spastic paraplegias • Dementias 	<ul style="list-style-type: none"> • Leigh syndrome • Neural tube defects • X-linked adrenoleukodystrophy
Prenatal genetics and fertility	<ul style="list-style-type: none"> • Abnormal antenatal scan findings • Abnormal antenatal test result • Non invasive prenatal testing and diagnosis • Family history of known genetic condition • Foetal exposure to teratogens • Previous foetal loss • Preimplantation genetic diagnosis • Genetic aspects of infertility • Foetal post mortem • Increased nuchal fold 	<ul style="list-style-type: none"> • Trisomy 13,18,21 • Sex chromosomal aneuploidy • Chromosome translocations and inversions • Supernumerary marker chromosomes • Hydrops fetalis • Exomphalos • Diaphragmatic hernia • Talipes
Dysmorphology	<ul style="list-style-type: none"> • Facial Dysmorphism • Overgrowth • Segmental overgrowth • Hemihypertrophy • Obesity • Growth retardation • Short Stature 	<ul style="list-style-type: none"> • Russell-Silver syndrome • Beckwith Wiedemann syndrome • Sotos syndrome • Noonan syndrome • Williams syndrome

	<ul style="list-style-type: none"> • Skeletal dysplasia • Limb anomalies • Developmental eye anomalies • Renal tract anomalies • Multiple Congenital malformations • Craniosynostosis • Cleft lip/palate • Craniofacial malformations • Ambiguous genitalia • Skin pigmentary abnormalities • Vascular Malformations • Arthrogryposis • Imprinting disorders 	<ul style="list-style-type: none"> • Kabuki syndrome • ARID1B/Coffin-Siris syndrome • De Lange syndrome • KBG syndrome • Neurofibromatosis type 1 • Tuberous sclerosis • Achondroplasia • Osteogenesis Imperfecta • Fragile X syndrome • DDX3X • SYNGAP1
Mitochondrial genetics		<ul style="list-style-type: none"> • Chronic progressive external ophthalmoplegia • Kearns-Sayre syndrome • Leber hereditary optic neuropathy • Leigh syndrome • MELAS • MERRF • NARP • Pearson syndrome
Metabolic Genetics		<ul style="list-style-type: none"> • Congenital Disorders of glycosylation • Fatty acid oxidation abnormalities • Glycogen storage diseases • Mucopolysaccharide disorders • Lysosomal storage diseases • Leukodystrophies • Organic acidemias • Neuronal ceroid lipofuscinoses • Peroxisomal disorders

4. Learning and Teaching

4.1 The training programme

The organisation and delivery of postgraduate training is the responsibility of the Health Education England (HEE), NHS Education for Scotland (NES), Health Education and Improvement Wales (HEIW) and the Northern Ireland Medical and Dental Training Agency (NIMDTA) – referred to from this point as ‘deaneries’. A training programme director (TPD) will be responsible for coordinating the specialty training programme. In England, the local organisation and delivery of training is overseen by a school of medicine.

Progression through the programme will be determined by the Annual Review of Competency Progression (ARCP) process and the training requirements for each indicative year of training are summarised in the ARCP decision aid (available on the [JRCPTB website](#)).

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 curriculum requirements are met and also that unnecessary duplication and educationally unrewarding experiences are avoided.

The following provides a guide on how training programmes should be focused in each training year in order for trainees to gain the experience and develop the capabilities to the level required.

Trainees will have an appropriate clinical supervisor and a named educational supervisor. The clinical supervisor and educational supervisor may be the same person.

Variant Interpretation

Trainees will need to gain appropriate expertise in genomic variant interpretation in order to achieve the speciality specific CiPs and to practice as a consultant clinical geneticist. Trainees can gain these skills by undertaking appropriate modules of a genomics MSc, as offered by many UK universities, or a PGcert in genomics as offered by St Georges hospital, London. Any trainee who is unable to pursue one of these routes will be able to plan a programme of learning with their Educational Supervisor which may for instance consist of attending an alternative variant interpretation course such as that offered by Wellcome Genome Courses and Conferences and combine this with practical experience gained by an appropriate period of working with senior scientists in one of the National Genomics Laboratories.

Counselling skills

Effective counselling skills are a crucial skill for doctors practicing in many medical disciplines, but the concepts to be discussed with patients can be particularly complex in Clinical Genetics and the potential consequences of a patient failing to understand a

disorder particularly far reaching. Trainees will be strongly encouraged to attend formal training in counselling skills such as that available via the UK universities genomics MSc's, or a similar course as deemed appropriate by the local Programme Director. Some UK training centres may have specific expertise in counselling theory and practice, and trainees in these centres may be able to acquire appropriate skills from local departmental teaching combined with selected clinical experience.

4.2 Teaching and learning methods

The curriculum will be delivered through a variety of learning experiences and will achieve the capabilities described in the syllabus 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.

This section identifies the types of situations in which a trainee will learn.

Workbased experiential learning - Trainees will undertake their learning in one or more regional genetics centres including all approved training hospitals associated that region, all relevant laboratories associated with that region and will include out of region experience for some specialist areas.

Medical clinics including specialty clinics

The educational objectives of attending clinics are:

- To understand the management of chronic diseases.
- Be able to assess a patient in a defined timeframe.
- To interpret and act on the referral letter to clinic.
- To propose an investigation and management plan in a setting different from the acute medical situation.
- Interpret genomic results in clinical context.
- To review and amend existing investigation plans.
- To write an acceptable letter back to the referrer.
- To communicate with the patient and where necessary relatives and other health care professionals.

These objectives can be achieved in a variety of settings including hospitals, day care facilities and the community. The clinic might be primarily run by a specialist nurse (or other qualified health care professionals) rather than a consultant physician. After initial induction, trainees will review patients in clinic settings, under direct supervision. The degree of responsibility taken by the trainee will increase as competency increases. Trainees should see a range of new and follow-up patients and present their findings to their clinical supervisor. Clinic letters written by the trainee should also be reviewed and feedback given.

The number of patients that a trainee should see in each clinic is not defined, neither is the time that should be spent in clinic, but as a guide this should be a minimum of two hours.

Clinic experience should be used as an opportunity to undertake supervised learning events and reflection.

Reviewing patients with consultants

It is important that trainees have an opportunity to present at least a proportion of the patients whom they have assessed to their consultant for senior review in order to obtain immediate feedback into their performance (that may be supplemented by an appropriate WBA such as a mini-CEX or CBD). This may be accomplished following a clinic undertaken with a consultant, or on a ward round with a consultant.

Personal ward rounds

Every patient seen, on the ward or in outpatients, provides a learning opportunity, which will be enhanced by following the patient through the course of their illness. The experience of the evolution of patients' problems over time is a critical part both of the diagnostic process as well as management. Patients seen should provide the basis for critical reading and reflection on clinical problems.

Ward rounds by more senior doctors

Every time a trainee observes another doctor seeing a patient or their relatives there is an opportunity for learning. Ward rounds should be led by a more senior doctor and include feedback on clinical and decision-making skills.

Multidisciplinary team meetings

There are many situations where clinical problems are discussed with clinicians in other disciplines. These provide excellent opportunities for observation of clinical reasoning.

As genomics increasingly underpins the understanding and management of many different areas of medical practice MDT working will provide invaluable learning for trainees, both in the genomics of those specialist areas of medicine, but also in the communication skills necessary for effective working in the modern NHS and negotiating skills vital for shared decision making.

MDT working with laboratory colleagues will also allow trainees to improve their skills interrogating and interpreting genomic data. Due to the nature of practicing in rare disease trainees may need to access information from a variety of sources in order to make a balanced judgement regarding a given clinical situation. This may include experts from other disciplines, and disease specific experts in clinical and laboratory arenas. Trainees will spend significant periods of time discussing clinical cases with senior colleagues which provides an important element of learning. Some of these sessions will be recorded on ePortfolio as formative learning events.

Laboratory Based Learning

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. Trainees will also spend time interrogating genomic data with laboratory and clinical colleagues and learning variant interpretation skills.

Academic Research

Many results generated by broad genomic testing may reveal genetic variation for which our current level of understanding is only partial. Trainees may become involved in research initiatives in order to further understand such variants or may initiate further laboratory testing to provide further clarity for a family or individual. Much of this work is on the cusp of our current understanding and requires research acumen to successfully progress forward. Trainees will improve their critical thinking around such research questions- which may require extensive literature reviews and liaison with national and international teams of experts. Many trainees will undertake small research projects in order to further refine these interpretation skills which may involve short periods of laboratory work or detailed investigation of a given clinical phenotype. Such undertakings can often be incorporated into the training programme.

In a discipline such as clinical genetics some trainees may wish to acquire additional research competencies (in addition to those required by the curriculum) and may consider applying for an OOPR and research funding in order to undertake a PhD or MD.

Formal postgraduate teaching

The content of these sessions is 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.

Suggested activities include:

- A programme of formal regular teaching sessions to cohorts of trainees (eg a weekly training hour).
- Case presentations, locally regionally and Nationally.
- Research, audit and quality improvement projects.
- Lectures and small group teaching.
- Grand Rounds.
- 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.
- National training courses covering specific aspects of the curriculum.
- Modules of approved courses leading to a PG Cert in genomics.

Training programme directors will advise how many regional and National meetings are required to gain capabilities depending on prior experience and local learning opportunities. As many areas of practice are very specialised in clinical genetics this balance may vary from one training centre to another.

Learning with peers - There are many opportunities for trainees to learn with their peers. Local postgraduate teaching opportunities allow trainees of varied levels of experience to come together for small group sessions.

Independent self-directed learning

Trainees entering clinical genetics are likely to be interested in basic science and independent self-directed learning is likely to be a relatively large part of practice. Trainees will use this time in a variety of ways depending upon their stage of learning.

Suggested activities include:

- reading, including web-based material such as e-Learning for Healthcare (e-LfH) and genomics packages
- maintenance of personal portfolio (self-assessment, reflective learning, personal development plan)
- audit, quality improvement and research projects
- reading journals
- achieving personal learning goals beyond the essential, core curriculum
- undertaking small research projects either clinical or laboratory based.

Formal study courses

Time to be made available for formal courses is encouraged, subject to local conditions of service. Examples include management and leadership courses and communication courses, which are particularly relevant to patient safety and experience.

4.3 Academic training

The four nations have different arrangements for academic training and doctors in training should consult the local deanery for further guidance.

Trainees may train in academic medicine as an academic clinical fellow (ACF), academic clinical lecturer (ACL) or equivalent.

Some trainees may opt to do research leading to a higher degree without being appointed to a formal academic programme. This new curriculum should not impact in any way on the facility to take time out of programme for research (OOPR) but as now, such time requires discussion between the trainee, the TPD and the Deanery as to what is appropriate together with guidance from the appropriate SAC that the proposed period and scope of study is sensible.

4.4 Taking time out of programme

There are a number of circumstances when a trainee may seek to spend some time out of specialty training, such as undertaking a period of research or taking up a fellowship post. All such requests must be agreed by the postgraduate dean in advance and trainees are advised to discuss their proposals as early as possible. Full guidance on taking time out of programme can be found in the Gold Guide.

4.5 Acting up as a consultant

A trainee coming towards the end of their training may spend up to three months “acting-up” as a consultant, provided that a consultant supervisor is identified for the post and satisfactory progress is made. As long as the trainee remains within an approved training programme, the GMC does not need to approve this period of “acting up” and their original CCT date will not be affected. More information on acting up as a consultant can be found in the Gold Guide.

5. Programme of Assessment

5.1 Purpose of assessment

The purpose of the programme of assessment is to:

- Assess trainees’ actual performance in the workplace.
- Enhance learning by providing formative assessment, enabling trainees to receive immediate feedback, understand 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.
- Demonstrate trainees have acquired the GPCs and meet the requirements of GMP
- Ensure that trainees possess the essential underlying knowledge required for their specialty.
- Provide robust, summative evidence that trainees are meeting the curriculum standards during the training programme.
- Inform the 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.

5.2 Programme of Assessment

Our programme of assessment refers to the integrated framework of exams, assessments in the workplace and judgements made about a learner during their approved programme of training. The purpose of the programme of assessment is to robustly evidence, ensure and clearly communicate the expected levels of performance at critical progression points in, and to demonstrate satisfactory completion of training as required by the curriculum.

The programme of assessment is comprised of several different individual types of assessment. A range of assessments is needed to generate the necessary evidence required for global judgements to be made about satisfactory performance, progression in, and completion of, training. All assessments, including those conducted in the workplace, are linked to the relevant curricular learning outcomes (eg through the blueprinting of assessment system to the stated curricular outcomes).

The programme of assessment emphasises the importance and centrality of professional judgement in making sure learners have met the learning outcomes and expected levels of performance set out in the approved curricula. Assessors will make accountable, professional judgements. The programme of assessment includes how professional judgements are used and collated to support decisions on progression and satisfactory completion of training.

The assessments will be supported by structured feedback for trainees. Assessment tools will be both formative and summative and have been selected on the basis of their fitness for purpose.

Assessment will take place throughout the training programme to allow trainees continually to gather evidence of learning and to provide formative feedback. Those assessment tools which are not identified individually as summative will contribute to summative judgements about a trainee's progress as part of the programme of assessment. 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.

Reflection and feedback should be an integral component to all SLEs and WBPAs. In order for trainees to maximise benefit, reflection and feedback should take place as soon as possible after an event. Every clinical encounter can provide a unique opportunity for reflection and feedback and this process should occur frequently. Feedback should be of high quality and should include an action plan for future development for the trainee. Both trainees and trainers should recognise and respect cultural differences when giving and receiving feedback.

5.3 Assessment of CiPs

Assessment of CiPs involves looking across a range of different skills and behaviours to make global decisions about a learner's suitability to take on particular responsibilities or tasks.

Clinical supervisors and others contributing to assessment will provide formative feedback to the trainee on their performance throughout the training year. This feedback will include a global rating in order to indicate to the trainee and their educational supervisor how they are progressing at that stage of training. To support this, workplace based assessments and multiple consultant reports will include global assessment anchor statements.

Global assessment anchor statements

- Below expectations for this year of training; may not meet the requirements for critical progression point
- Meeting expectations for this year of training; expected to progress to next stage of training
- Above expectations for this year of training; expected to progress to next stage of training

Towards the end of the training year, trainees will make a self-assessment of their progression for each CiP and record this in the ePortfolio with signposting to the evidence to support their rating.

The educational supervisor (ES) will review the evidence in the ePortfolio including workplace based assessments, feedback received from clinical supervisors (via the Multiple Consultant Report) and the trainee's self-assessment and record their judgement on the trainee's performance in the ES report, with commentary.

For **generic CiPs**, the ES will indicate whether the trainee is meeting expectations or not using the global anchor statements above. Trainees will need to be meeting expectations for the stage of training as a minimum to be judged satisfactory to progress to the next training year.

For **specialty CiPs**, the ES will make an entrustment decision for each CiP and record the indicative level of supervision required with detailed comments to justify their entrustment decision. The ES will also indicate the most appropriate global anchor statement (see above) for overall performance.

Level descriptors for specialty CiPs

Level	Descriptor
Level 1	Entrusted to observe only – no provision of clinical care
Level 2	Entrusted to act with direct supervision: The trainee may provide clinical care, but the supervising physician is physically within the hospital or other site of patient care and is immediately available if required to provide direct bedside supervision
Level 3	Entrusted to act with indirect supervision: The trainee may provide clinical care when the supervising physician is not physically present within the hospital or other site of patient care, but is

	available by means of telephone and/or electronic media to provide advice, and can attend at the bedside if required to provide direct supervision
Level 4	Entrusted to act unsupervised

The ARCP will be informed by the ES report and the evidence presented in the ePortfolio. The ARCP panel will make the final summative judgement on whether the trainee has achieved the generic outcomes and the appropriate level of supervision for each CiP. The ARCP panel will determine whether the trainee can progress to the next year/level of training in accordance with the Gold Guide. ARCPs will be held for each training year. The final ARCP will ensure trainees have achieved level 4 in all CiPs for the critical progression point at completion of training.

5.4 Critical progression points

There will be a key progression point on completion of specialty training. Trainees will be required to be entrusted at level 4 in all CiPs by the end of training in order to achieve an ARCP outcome 6 and be recommended for a CCT.

The educational supervisor report will make a recommendation to the ARCP panel as to whether the trainee has met the defined levels for the CiPs and acquired the procedural competence required for each year of training. The ARCP panel will make the final decision on whether the trainee can be signed off and progress to the next year/level of training [see section 5.6].

The outline grid below sets out the expected level of supervision and entrustment for the specialty CiPs and includes the critical progression point at the end of the training programme.

Table 1: Outline grid of levels expected for Clinical Genetics specialty CiPs

Level descriptors

Level 1: Entrusted to observe only – no clinical care

Level 2: Entrusted to act with direct supervision

Level 3: Entrusted to act with indirect supervision

Level 4: Entrusted to act unsupervised

Specialty CiP	Specialty training				CCT
	ST3	ST4	ST5	ST6	CRITICAL PROGRESSION POINT
1. Managing a comprehensive genetic medicine service for both inpatients and outpatients	2	2	3	4	
2.. Working within multidisciplinary teams and consultations related to the management and treatment of complex genetic disorders	1	2	2	4	
3. Managing predictive genetic testing and advising on cascade genetic testing in families.	2	3	3	4	
4. Managing storage and testing of genetic material in the prenatal and post mortem settings.	2	2	3	4	
5. Interrogating and interpreting genetic data and communicating effectively with laboratory colleagues.	1	2	3	4	
6. Contributing to genetic research and clinical trials.	2	2	2	4	

5.5 Evidence of progress

The following methods of assessment will provide evidence of progress in the integrated programme of assessment. The requirements for each training year/level are stipulated in the ARCP decision aid (www.jrcptb.org.uk).

Summative assessment

Examinations and certificates

- KBA Certificate Examination in Medical Genetics

The Royal College of Pathologists provides the Certificate Examination in Medical Genetics and further information including details of the format of the exam can be found on the webpage: <https://www.rcpath.org/trainees/examinations/examinations-by-specialty/medical-genetics.html>

Formative assessment

Supervised Learning Events (SLEs)

- Case-Based Discussions (CbD)
- mini-Clinical Evaluation Exercise (mini-CEX)

WPBA

- Multi-Source Feedback (MSF)
- Patient Survey (PS)
- Quality Improvement Project Assessment Tool (QIPAT)
- Teaching Observation (TO)

Supervisor reports

- Multiple Consultant Report (MCR)
- Educational Supervisor Report (ESR)

These methods are described briefly below. More information and guidance for trainees and assessors are available in the ePortfolio and on the JRCPTB website (www.jrcptb.org.uk).

Assessment should be recorded in the trainee's ePortfolio. These methods include feedback opportunities as an integral part of the programme of assessment.

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 focus on 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.

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.

Multi-source 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 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, administrative staff, and other allied professionals. Raters should be agreed with the educational supervisor at the start of the training year. The trainee will not see the individual responses by raters. Feedback is given to the trainee by the Educational Supervisor.

Patient Survey (PS)

A trainee's interaction with patients should be continually observed and assessed. The Patient Survey provides a tool to assess a trainee during a consultation period. The Patient Survey assesses the trainee's performance in areas such as interpersonal skills, communication skills and professionalism.

Quality Improvement Project Assessment Tool (QIPAT)

The QIPAT is designed to assess a trainee's competence in completing a quality improvement project. The QIPAT can be based on review of quality improvement project documentation or on a presentation of the quality improvement project at a meeting. If possible, the trainee should be assessed on the same quality improvement project by more than one assessor.

Teaching Observation (TO)

The TO form is designed to provide structured, formative feedback to trainees on their competence at teaching. The TO 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).

Multiple Consultant Report (MCR)

The MCR captures the views of consultant supervisors based on observation on a trainee's performance in practice. The MCR feedback and comments received give valuable insight into how well the trainee is performing, highlighting areas of excellence and areas of support required. MCR feedback will be available to the trainee and contribute to the educational supervisor's report.

Educational supervisors report (ESR)

The ES will periodically (at least annually) record a longitudinal, global report of a trainee's progress based on a range of assessment, potentially including observations in practice or reflection on behaviour by those who have appropriate expertise and experience. The ESR will include the ES's summative judgement of the trainee's performance and the entrustment decisions given for the learning outcomes (CiPs). The ESR can incorporate commentary or reports from longitudinal observations, such as from supervisors (MCRs) and formative assessments demonstrating progress over time.

5.6 Decisions on progress (ARCP)

The decisions made at critical progression points and upon completion of training should be clear and defensible. They must be fair and robust and make use of evidence from a range of assessments, potentially including exams and observations in practice or reflection on behaviour by those who have appropriate expertise or experience. They can also incorporate commentary or reports from longitudinal observations, such as from supervisors or formative assessments demonstrating progress over time.

Periodic (at least annual) review should be used to collate and systematically review evidence about a doctor's performance and progress in a holistic way and make decisions about their progression in training. The annual review of progression (ARCP) process supports the collation and integration of evidence to make decisions about the achievement of expected outcomes.

Assessment of CiPs involves looking across a range of different skills and behaviours to make global decisions about a learner's suitability to take on particular responsibilities or tasks, as do decisions about the satisfactory completion of presentations/conditions and procedural skills set out in this curriculum. The outline grid in section 5.4 sets out the level of supervision expected for each of the clinical and specialty CiPs. The requirements for each year of training are set out in the ARCP decision aid (www.jrcptb.org.uk).

The ARCP process is described in the Gold Guide. Deaneries are responsible for organising and conducting ARCPs. The evidence to be reviewed by ARCP panels should be collected in the trainee's ePortfolio.

As a precursor to ARCPs, JRCPTB strongly recommend that trainees have an informal ePortfolio review either with their educational supervisor or arranged by the local school of

medicine. These provide opportunities for early detection of trainees who are failing to gather the required evidence for ARCP.

There should be review of the trainee’s progress to identify any outstanding targets that the trainee will need to complete to meet all the learning outcomes for completion training approximately 12-18 months before CCT. This should include an external assessor from outside the training programme.

In order to guide trainees, supervisors and the ARCP panel, JRCPTB has produced an ARCP decision aid which sets out the requirements for a satisfactory ARCP outcome at the end of each training year and critical progression point. The ARCP decision aid is available on the JRCPTB website www.jrcptb.org.uk.

Poor performance should be managed in line with the Gold Guide.

5.7 Assessment blueprint

The table below show the possible methods of assessment for each CiP. It is not expected that every method will be used for each competency and additional evidence may be used to help make a judgement on capability.

KEY

DOPS	Direct observation of procedural skills	CbD	Case-based discussion
MCR	Multiple consultant report	MiniCEX	Mini-clinical evaluation exercise
PS	Patient survey	MSF	Multi source feedback
TO	Teaching observation	QIPAT	Quality improvement project assessment tool
KBA	Certificate Examination in Medical Genetics		

Blueprint for WPBAs mapped to CiPs

Learning outcomes	ACAT	CbD	DOPS	MCR	Mini-CEX	MSF	PS	QIPAT	TO	KBA
Generic CiPs										

Able to function successfully within NHS organisational and management systems				√		√				
Able to deal with ethical and legal issues related to clinical practice		√	√	√	√	√				
Communicates effectively and is able to share decision making, while maintaining appropriate situational awareness, professional behaviour and professional judgement				√		√	√			
Is focused on patient safety and delivers effective quality improvement in patient care				√		√		√		
Carrying out research and managing data appropriately				√		√				
Acting as a clinical teacher and clinical supervisor				√		√			√	
Specialty CiPs										
Managing a comprehensive genetic medicine service for both inpatients and outpatients.		√		√	√	√	√			√
Working within multidisciplinary teams and consultations related to the management and treatment of complex genetic disorders.		√		√		√				√
Managing predictive genetic testing and advising on cascade genetic testing in families.		√		√	√	√	√			
Managing storage and testing of genetic material in the prenatal and post mortem settings.		√		√		√		√		
Interrogating and interpreting genetic data and communicating effectively with laboratory colleagues.		√		√		√		√		√
Contributing to genetic research and clinical trials.		√		√				√		√

6. Supervision and feedback

This section of the curriculum describes how trainees will be supervised, and how they will receive feedback on performance. For further information please refer to the AoMRC guidance on Improving feedback and reflection to improve learning⁵.

Access to high quality, supportive and constructive feedback is essential for the professional development of the trainee. Trainee reflection is an important part of the feedback process and exploration of that reflection with the trainer should ideally be a two-way dialogue. Effective feedback is known to enhance learning and combining self-reflection to feedback promotes deeper learning.

Trainers should be supported to deliver valuable and high quality feedback. This can be by providing face to face training to trainers. Trainees would also benefit from such training as they frequently act as assessors to junior doctors, and all involved could also be shown how best to carry out and record reflection.

6.1 Supervision

All elements of work in training posts must be supervised with the level of supervision varying depending on the experience of the trainee and the clinical exposure and case mix undertaken. Outpatient and referral supervision must routinely include the opportunity to discuss all cases with a supervisor if appropriate. As training progresses the trainee should have the opportunity for increasing autonomy, consistent with safe and effective care for the patient.

Organisations must make sure that each doctor in training has access to a named clinical supervisor and a named educational supervisor. Depending on local arrangements these roles may be combined into a single role of educational supervisor. However, it is preferred that a trainee has a single named educational supervisor for (at least) a full training year, in which case the clinical supervisor is likely to be a different consultant during some placements.

The role and responsibilities of supervisors have been defined by the GMC in their standards for medical education and training⁶.

Educational supervisor

⁵ [Improving feedback and reflection to improve learning. A practical guide for trainees and trainers](#)

⁶ [Promoting excellence: standards for medical education and training](#)

The educational supervisor is responsible for the overall supervision and management of a doctor's educational progress during a placement or a series of placements. The educational supervisor regularly meets with the doctor in training to help plan their training, review progress and achieve agreed learning outcomes. The educational supervisor is responsible for the educational agreement, and for bringing together all relevant evidence to form a summative judgement about progression at the end of the placement or a series of placements.

Clinical supervisor

Consultants responsible for patients that a trainee looks after provide clinical supervision for that trainee and thereby contribute to their training; they may also contribute to assessment of their performance by completing a 'Multiple Consultant Report (MCR)' and other WPBAs. A trainee may also be allocated (for instance, if they are not working with their educational supervisor in a particular placement) a named clinical supervisor, who is responsible for reviewing the trainee's training and progress during a particular placement. It is expected that a named clinical supervisor will provide a MCR for the trainee to inform the Educational Supervisor's report.

The educational and (if relevant) clinical supervisors, when meeting with the trainee, should discuss issues of clinical governance, risk management and any report of any untoward clinical incidents involving the trainee. If the service lead (clinical director) has any concerns about the performance of the trainee, or there are issues of doctor or patient safety, these would be discussed with the clinical and educational supervisors (as well as the trainee). 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.

Educational and clinical supervisors need to be formally recognised by the GMC to carry out their roles⁷. It is essential that training in assessment is provided for trainers and trainees in order to ensure that there is complete understanding of the assessment system, assessment methods, their purposes and use. Training will ensure a shared understanding and a consistency in the use of the WPBAs and the application of standards.

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.

Trainees

Trainees should make the safety of patients their first priority and they should not be practising in clinical scenarios which are beyond their experiences and competencies without supervision. Trainees should actively devise individual learning goals in discussion with their trainers and should subsequently identify the appropriate opportunities to achieve said learning goals. Trainees would need to plan their WPBAs accordingly to enable

⁷ [Recognition and approval of trainers](#)

their WPBAs to collectively provide a picture of their development during a training period. Trainees should actively seek guidance from their trainers in order to identify the appropriate learning opportunities and plan the appropriate frequencies and types of WPBAs according to their individual learning needs. It is the responsibility of trainees to seek feedback following learning opportunities and WPBAs. Trainees should self-reflect and self-evaluate regularly with the aid of feedback. Furthermore, trainees should formulate action plans with further learning goals in discussion with their trainers.

6.2 Appraisal

A formal process of appraisals and reviews underpins training. This process ensures adequate supervision during training, 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 e-portfolio at this time, recording their commitment to the training process.

Mid-point Review

This meeting between trainee and educational supervisor is not mandatory (particularly 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 ePortfolio. 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 e-portfolio. 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. Supervisors should also identify areas where a trainee has performed about the level expected and highlight successes.

7. Quality Management

The organisation of training programs is the responsibility of the deaneries. The deaneries will oversee programmes for postgraduate medical training in their regions. The Schools of Medicine in England, Wales and Northern Ireland and the Medical Specialty Training Board in Scotland will undertake the following roles:

- oversee recruitment and induction of trainees into the specialty
- allocate trainees into particular rotations appropriate to their training needs
- oversee the quality of training posts provided locally
- ensure adequate provision of appropriate educational events □ ensure curricula implementation across training programmes
- oversee the workplace-based assessment process within programmes
- coordinate the ARCP process for trainees
- provide adequate and appropriate career advice
- provide systems to identify and assist doctors with training difficulties □ provide flexible training.

Educational programmes to train educational supervisors and assessors in workplace based assessment may be delivered by deaneries or by the colleges or both.

Development, implementation, monitoring and review of the curriculum are the responsibility of the JRCPTB and the SAC. The committee will be formally constituted with representatives from each health region in England, from the devolved nations and with trainee and lay representation. It will be the responsibility of the JRCPTB to ensure that curriculum developments are communicated to heads of school, regional specialty training committees and TPDs.

The JRCPTB has a role in quality management by monitoring and driving improvement in the standard of all medical specialties on behalf of the three Royal Colleges of Physicians in Edinburgh, Glasgow and London. The SACs are actively involved in assisting and supporting deaneries to manage and improve the quality of education within each of their approved training locations. They are tasked with activities central to assuring the quality of medical education such as writing the curriculum and assessment systems, reviewing applications for new posts and programmes, provision of external advisors to deaneries and recommending trainees eligible for CCT or Certificate of Eligibility for Specialist Registration (CESR).

JRCPTB uses data from six quality datasets across its specialties and subspecialties to provide meaningful quality management. The datasets include the GMC national Training Survey (NTS) data, ARCP outcomes, examination outcomes, new consultant survey, penultimate year assessments (PYA)/external advisor reports and the monitoring visit reports.

Quality criteria have been developed to drive up the quality of training environments and ultimately improve patient safety and experience. These are monitored and reviewed by JRCPTB to improve the provision of training and ensure enhanced educational experiences.

8. Intended use of curriculum by trainers and trainees

This curriculum and ARCP decision aid are available from the Joint Royal Colleges of Physicians Training Board (JRCPTB) via the website www.jrcptb.org.uk.

Clinical and educational supervisors should use the curriculum and decision aid as the basis of their discussion with trainees, particularly during the appraisal process. 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 an ePortfolio. The trainee will use the curriculum to develop learning objectives and reflect on learning experiences.

Recording progress in the ePortfolio

On enrolling with JRCPTB trainees will be given access to the ePortfolio which 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 it 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 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.

Deaneries, training programme directors, college tutors and ARCP panels may use it to monitor the progress of trainees for whom they are responsible.

JRCPTB will use summarised, anonymous ePortfolio data to support its work in quality assurance.

All appraisal meetings, personal development plans and workplace based assessments (including MSF) should be recorded in the ePortfolio. Trainees are encouraged to reflect on their learning experiences and to record these in the ePortfolio. Reflections can be kept private or shared with supervisors.

Reflections, assessments and other ePortfolio content should be used to provide evidence towards acquisition of curriculum capabilities. Trainees should add their own self-assessment ratings to record their view of their progress. The aims of the self-assessment are:

- To provide the means for reflection and evaluation of current practice.
- To inform discussions with supervisors to help both gain insight and assists in developing personal development plans.
- To identify shortcomings between experience, competency and areas defined in the curriculum so as to guide future clinical exposure and learning.

Supervisors can sign-off and comment on curriculum capabilities to build up a picture of progression and to inform ARCP panels.

9. Equality and diversity

The Royal Colleges of Physicians will comply, and ensure compliance, with the requirements of equality and diversity legislation set out in the Equality Act 2010.

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.

Deaneries quality assurance will ensure that each training programme complies with the equality and diversity standards in postgraduate medical training as set by GMC. They should provide access to a professional support unit or equivalent for trainees requiring additional support.

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 ensuring that educational supervisors have had equality and diversity training (for example, an e-learning module) every three years.
- Deaneries ensuring that any specialist participating in trainee interview/appointments committees or processes has had equality and diversity training (at least as an e-module) every three 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.

- Providing resources to trainees needing support (for example, through the provision of a professional support unit or equivalent).
- Monitoring of college Examinations.
- Ensuring all assessments discriminate on objective and appropriate criteria and do not unfairly advantage or disadvantage a trainee with any of the Equality Act 2010 protected characteristics. All efforts shall be made to ensure the participation of people with a disability in training through reasonable adjustments.

JRCPTB

Joint Royal Colleges of Physicians Training Board

