INSTITUTE
OF MEDICINE

ROYAL COLLEGE OF
PHYSICIANS OF IRELAND

HIGHER SPECIALIST TRAINING IN

CLINICAL GENETICS

OUTCOME BASED EDUCATION CURRICULUM

This Curriculum of Higher Specialist Training in Clinical Genetics was developed in 2023 The Curriculum undergoes an annual review process by the National Specialty Director(s) and the RCPI Education Department. The Curriculum is approved by the Specialty Training Committee and the Institute of Medicine.

Version	Date Published	Last Edited By	Version Comments
2.0	July 2026	Keith Farrington	Changes to specialty specific course content. Updates to the Core Professional Skills section to explicitly align with the *Eight Domains of Good Professional Practice*.

National Specialty Director's Foreword

This curriculum is intended to guide learning and provide a road map for trainees in Clinical Genetics. The Clinical Genetics HST Programme aims to deliver expert Clinical Geneticists with a broad range of clinical and academic skills.

This curriculum is designed to produce well-rounded graduates with the ability to manage the genetic care of clinical genetic conditions while supporting the development of subspecialty expertise and academic interests. The Outcome Based Education (OBE) project concerns the transition of the current minimum requirements model of the clinical genetics' curriculum and training across to OBE, which is more in line with other countries in Europe and the US. It is one of the key initiatives of the RCPI's Strategic Plan 2021 – 2024 which aims to enhance the quality of Ireland's BST and HST training programmes to ensure they are aligned with international best practices and standards. This will involve a considerable change to both the structure and assessment of the curriculum and as such requires input from multiple stakeholders to ensure that any changes are valid and robust. This RCPI Higher Specialist Training Outcome Based Education Curriculum provides the framework for the training of doctors that will produce excellent clinicians competent to practice as Consultant Clinical Geneticists.

The Clinical Genetics HST curriculum will now be an outcomes-based program. There are six broad training goals each aligned to key areas of practice. Within each goal are a series of training outcomes that reflect the sum of day-to-day practice in Clinical Genetics. Trainees will demonstrate proficiencies in each outcome matched to the level of their training, progressing to independent competence in each. Trainers will link closely with their trainees assisting them and evaluating their progress on a regular basis. In addition to clinical training, and reflecting the diversity of the specialty, the curriculum includes management, education, quality improvement, laboratory experience and opportunities for research experience. It highlights the increasing importance of case discussions, multidisciplinary meetings, and communication, especially acknowledging diagnostic and clinical uncertainty and reflecting the increasing complexity of the genetic conditions. I hope that this document will provide guidance, both for the trainees on this journey, and to trainers, to allow for meaningful dialogue, feedback, and support. The ultimate goal of this document is to enhance training and prepare future clinical leaders in Clinical Genetics.

I would like to thank all the trainers that took part in the workshop and discussion group that finalised the new curriculum. I am excited to be part of the ongoing development of the Clinical Genetics HST programme with trainers and colleagues in the RCPI. I wish all our Trainees good luck as they embark on their training and their clinical careers.

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INTRODUCTION

This section includes an overview of the Higher Specialist Training programme and of this Curriculum document.

Purpose of Training

This programme is designed to provide training in Clinical Genetics in approved training posts, under supervision, to fulfil agreed curricular requirements. Each post provides a trainee with a named trainer and the programme is under the direction of the National Specialty Director in Clinical Genetics.

Purpose of the Curriculum

The purpose of the Curriculum is to guide the Trainee towards achieving the educational outcomes necessary to work as an independent consultant. The Curriculum defines the relevant processes, content, outcomes, and requirements to be achieved. It stipulates the overarching goals, outcomes, expected learning experiences, instructional resources and assessments that comprise the Higher Specialist Training (HST) programme. It provides a framework for certifying successful completion of HST programme.

In keeping with developments in medical education and to ensure alignment with international best practice and standards, the Royal College of Physicians (RCPI) have implemented an Outcomes Based Education (OBE) approach. This curriculum design differs from traditional minimum based requirement designs in that the learning process and desired end-product of training (outcomes) are at the forefront of the design to provide the essential training opportunities and experiences to achieve those outcomes.

How to Use the Curriculum

Trainees and Trainers should use the Curriculum as a basis for goal-setting meetings, delivering feedback, and completing assessments, including appraisal processes (Quarterly Assessments/End of Post Assessment, End of Year Evaluation). Therefore, it is expected that both Trainees and Trainers familiarise themselves with the Curriculum and have a good working knowledge of it.

Trainees are expected to use the Curriculum as a blueprint for their training and record specific feedback, assessments, and training events on ePortfolio. The ePortfolio should be updated frequently during each training placement.

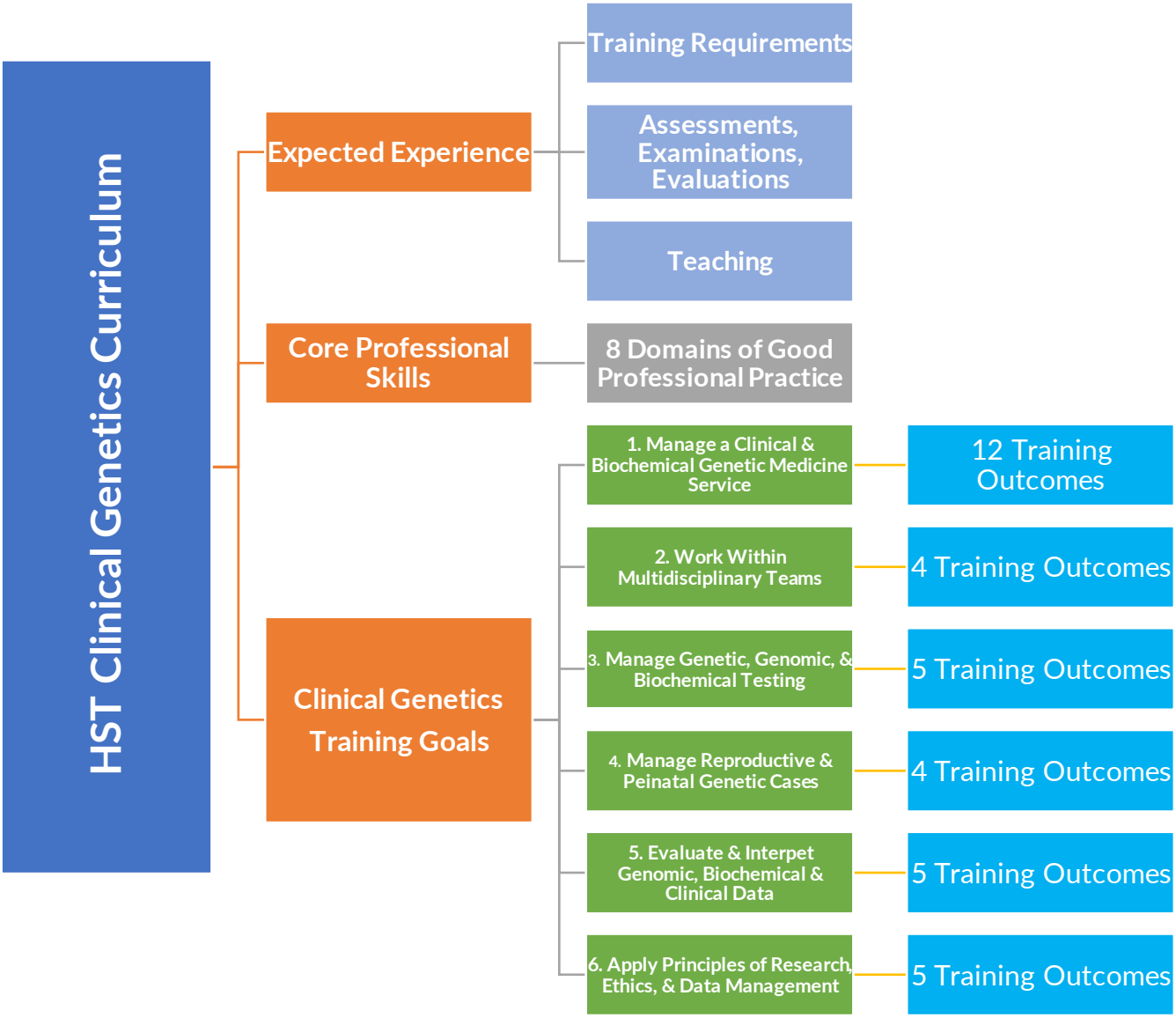
It is important to note that ePortfolio is a digital repository designed to reflect Curriculum requirements. It facilitates recording of progress through HST and evidence that training is valid and appropriate. While a complete ePortfolio is essential for HST certification, Trainees and Trainers should always refer to the Curriculum in the first instance for information on the requirements of the training programme.

Please note: It is the responsibility of the Trainee to keep an up-to-date ePortfolio throughout the programme as it reflects their individual training experience and it documents that they have successfully met training standards as expected by the Medical Council.

Reference to Rules & Regulations

Please refer to the following sections within the Clinical Genetics HST Training Handbook for rules and regulations associated with this post. Policies, procedures, relevant documents, and Training Handbooks can be accessed on the RCPI website following this link.

Overview of Curriculum Sections & Training Goals



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EXPECTED EXPERIENCE

This section details the training experience that all Trainees are expected to complete over the course of Higher Specialist Training.

Post Structure

The duration of HST in Clinical Genetics is four years. While all four years can be completed in HST training posts, trainees are encouraged to consider Out of Programme (OPE) training opportunities as part of their programme (see below).

Core Training

Trainees must spend the first two years of training in HST clinical posts in Ireland. The programme aims to be flexible in terms of sequence of training after this time. The first two years are directed towards acquiring a broad general experience of Clinical Genetics under appropriate supervision. An increase in the content of hands-on experience follows naturally and, as confidence is gained and abilities are acquired, the trainee will be encouraged to assume a greater degree of responsibility and independence.

Out of Clinical Programme Experience (OCPE)

Trainees can undertake one, or more years out of their HST programme to pursue research, further education, special clinical training, lecturing experience or other relevant experiences.

OCPE must be preapproved, and retrospective credit cannot be applied.

It must be noted that even if trainees can undertake more than one year to complete their OCPE of choice, RCPI would award a maximum of 12 months of training credits towards the achievement of CSCST. In certain circumstances, RCPI may award no credits. The decision of whether to award credits for one year may differ from specialty to specialty and it is discretionary by the NSDs of each respective specialty.

For more information on OCPE, please refer to the RCPI website (here).

Training Principles

During the period of training the Trainee must take increasing responsibility for seeing patients, undertaking ward consultations, making decisions and operating at a level of responsibility which would prepare them for practice as an independent Consultant. Over the course of HST, Trainees are expected to gain experience in a variety of hospital settings.

Core Professional Skills

Generic knowledge, skills and attitudes support competencies that are common to good medical practice in all of the medical and related specialties. It is intended that all Trainees should re-affirm those competencies during Higher Specialist Training. No timescale of acquisition is imposed, but failure to make progress towards meeting these important objectives at an early stage would cause concern about a Trainee's suitability and ability to become an independent specialist.

Recording of Evidence of Training

The target numbers for training items in the following sections represent the recording requirement to document evidence of relevant and varied clinical experience; it is understood that actual number of training experiences is likely to be well in excess of these numbers.

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To complete the HST Training Programme in Clinical Genetics, Trainees are expected to observe the following rotations requirements.

Over the course of HST, Trainees are expected to complete:

- 48 months (4 x 12 months) experience in Clinical Genetics Posts
- Rotation experience will be acquired in each of the centres in Ireland (Mater Misericordiae Hospital and CHI Crumlin.)
- At the start of each post, trainees are expected to fill out a Personal Goals form with their trainer and upload it on ePortfolio; the form should be agreed and signed by both Trainee & Trainer
- Specified Laboratory Experience – experience in biochemical genetics Laboratory, newborn screening laboratory, cytogenetics laboratory, molecular genetics laboratory, genomics laboratory.

Failure to demonstrate satisfactory progress at end of year review or in relation to examinations may result delay training or prevent its completion.

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Outpatient Clinics, Ward Rounds, Consultations, & Training Activities

Attendance at Clinics, participation in Ward Rounds and Patient Consultations are required elements of all posts throughout the programme. The timetable and frequency of attendance should be agreed with the assigned trainer at the beginning of the post.

This table provides an overview of the expected experience a Specialist Registrar should gain regarding clinics attendance, ward rounds, laboratory activities and consultations.

CLINICAL ACTIVITIES			
Clinic	**Timeline**	**Expected Experience**	**ePortfolio Form**
Biochemical Genetics	Years 1-4	Attend at least 1 per week in appropriate post	Clinics
General Genetics	Years 1-4	Attend at least 1 per week in appropriate post	Clinics
Cardiac Genetics	Years 1-4	20 per programme (in appropriate post)	Clinics
Adult Neurogenetics	Years 1-4	20 per programme (in appropriate post)	Clinics
Ophthalmology Genetics	Years 1-4	10 per programme (in appropriate post)	Clinics
Cystic Fibrosis	Years 1-4	Attend where appropriate	Clinics
Cancer	Years 1-4	Attend at least 1 per week in appropriate post	Clinics
Neurofibromatosis/Tuberous sclerosis	Years 1-4	Attend at least 1 per week	Clinics
Fetal Medicine	Years 1-4	Attend at least 1 per week in appropriate post	Clinics
CONSULTATIONS, MDT, PROCEDURES, LABORATORY			
Type	**Timeline**	**Expected Experience**	**ePortfolio Form**
Consultations	Years 1-4	At least 2 per month	Clinical Activities
MDT/ Meetings			
• Medicine (Cardiology)	Years 1-4	Monthly – to attend in appropriate post	Clinical Activities
• Paediatric cancer genetics (Crumlin)	Years 1-4	Monthly – to attend in appropriate post	Clinical Activities
• Cross City (Paediatrics) Neurology meeting	Years 1-4	Twice yearly – to attend in appropriate post	Clinical Activities
• Paediatric Disorders of Sexual Development (Endocrine)	Years 1-4	Twice yearly – to attend in appropriate post	Clinical Activities
• Dublin tri-hospital Fetal Medicine group	Years 1-4	Thrice yearly – to attend in appropriate post	Clinical Activities
• Variant interpretation meetings	Years 1-4	Every 2 months – to attend in appropriate post	Clinical Activities
Procedures			
• Skin biopsy	Years 1-4	As required	Clinical Activities
• Buccal Swab	Years 1-4	As required	Clinical Activities
Laboratory Experience	Years 1-4	As required	Clinical Activities
Examinations (either of)			
European Board of Medical Genetics	Years 1-4	1 Per Programme	Examinations
Royal College of Pathologists Certificate in Medical Genetics	Years 1-4	1 Per Programme	Examinations

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Hospital Based Learning & In-House Commitments

Specialist Registrars are expected to attend a series of in-house commitments as follows:

- Attend at least **1 Grand Rounds per month**
- Attend at least **1 Journal Club per month**
- Attend at least **1 MDT Meeting per week**
- Attend at least **1 Seminar, teaching session or journal club per month**
- Attend at least **1 Lecture / Webinar per quarter**

Research, Audit, & Teaching Experiences

Specialist Registrars are expected to complete the following activities:

- Deliver **12 teaching sessions** (to include tutorials, lectures, bedside teaching, etc.) over the course of 4 years of HST
- Deliver **1 oral presentation**, per each year of HST
- Complete **1 Audit or Quality Improvement Project**, per year of HST
- Attend **1 National or International Meeting**, per each year of HST
- Complete **1 research project**, over the course of 4 years of HST
- Complete **1 publication** (may include peer reviewed research, case report or patient information that demonstrates effective written communication or scientific writing,) over the course of 4 years of HST

Teaching Attendance

Specialist Registrars are expected to attend all the courses and study days as detailed in the Teaching Appendix, at the end of this document.

Assessments, Evaluations, & Examinations

Specialist Registrars are expected to:

- **4 quarterly assessments per training year** (1 assessment per quarter)
- **1 end of year evaluation at the end of each training year**
- **Regularly update your ePortfolio – this is your record of training and is a vital resource**
- **Complete all relevant workplace-based assessments in partnership with your trainer**
- Complete all the **workplace-based assessments** as outlined in the table below and as agreed with Trainer. It is recommended to **record at least 1 WBA** (CBD, MiniCEX, or DOPS) **per quarter** to be reviewed at the Quarterly Assessment.
- **Complete examination as advised by Trainer(s)**

For more information on evaluations, assessment, and examinations, please refer to the Assessment Appendix at the end of this document.

Summary of Expected Experience

Experience Type	Trainee is expected to	ePortfolio Form
Rotation Requirements	Complete all requirements related to the posts agreed	n/a
Personal Goals	At the start of each post complete a Personal Goals form on ePortfolio, agreed with your trainer and signed by both Trainee & Trainer	Personal Goals
Clinics	Attend outpatient Clinics as agreed with your trainer and record attendance per each post on ePortfolio	Clinics
Deliver Teaching	Record on ePortfolio all the occurrences where you have delivered Tutorials (at least 1 per Year), Lectures (at least 1 per Year), and Bedside or clinic teaching (at least 4 per Year)	Delivery of Teaching
Research	Desirable Experience: actively participate in research, seek to publish a paper and present research at conferences or national/international meetings	Research Activities
Publication	Complete 1 publication during the training programme	Additional Professional Activities
Presentation	Deliver 1 oral presentation or poster per each year of training	Additional Professional Activities
Audit	Complete and report on an audit or Quality Improvement (QI)per each year of training, either to start, continue or complete	Audit and QI
Attendance at In-House Activities	Attend at least 1 Grand Rounds per month, Attend at least 1 MDT Meeting (see above) per week, Attend at least 1 Seminar/Journal Club/Educational session per month, Attend at least 1 Lecture/Webinar per quarter Record attendance on ePortfolio	Attendance at In-House Activities
National/International Meetings	Attend 1 per year of training	Additional Professional Activities
Teaching Attendance	Attend courses and Study Days as detailed in the Teaching Appendix	Teaching Attendance
Examinations	European Board of Medical Genetics **or** Royal College of Pathologists Certificate in Medical Genetics	Examinations
Evaluations and Assessments	Complete a Quarterly Assessment/End of post assessment with your trainer 4 times in each year. Discuss your progress and complete the form.	Quarterly Assessments/End-of-Post Assessments
Workplace-based Assessment	Complete all the workplace-based assessment as agreed with your trainer and complete the respective form.	CBD/DOPS/Mini-CEX
End of Year Evaluation	Prepare for your End of Year Evaluation by ensuring your portfolio is up to date and your End of Year Evaluation form is initiated with your trainer.	End of Year Evaluation

CORE PROFESSIONAL SKILLS

This section includes the Irish Medical Council guidelines for medical professional conduct.

*The Medical Council has defined **eight domains of good professional practice**.*

These domains describe a framework of competencies applicable to all doctors across the continuum of professional development from formal medical education and training through to maintenance of professional competence. They describe the outcomes which doctors should strive to achieve and doctors should refer to these domains throughout the process of maintaining competence.

These principles are woven into training practice and feedback is formally provided in the Quarterly Assessments, End of Post, and End of Year Evaluation.

Core Professional Skills

The Core Professional Skills (CPS), updated in 2026, define the standards of professional practice expected of all doctors in postgraduate training across RCPI specialties. Aligned with the *Eight Domains of Good Professional Practice* (**Irish Medical Council, 2024**), they provide the standards of practice across the continuum of professional development, from formal medical education and training through the maintenance of professional competence.

The CPS are embedded within the formal structures of training and are developed through clinical practice, workplace-based learning, supervision, and structured educational activity. CPS are assessed through workplace-based assessment, quarterly assessments, and ePortfolio evidence, ensuring that Trainees embed professionalism in their practice. Within this, the RCPI Taught Programme provides structured educational content through which CPS standards are addressed and contextualised across all levels of training.

Within each domain identified by the Medical Council, the cross-speciality RCPI Clinical Working Group has articulated key areas of professional practice and relevant expectations for training. Collectively, the domains support professionalism as a core component of safe, effective, and patient-centred care.

1 Patient Safety and Quality of Patient Care

Doctors must place patient safety and quality of care at the centre of practice, ensuring accountability to patients, their profession, and their organisation. This requires addressing risks, managing incidents, preventing infection, and driving continuous improvement within governance and ethical standards. By embedding safety and accountability into practice, doctors protect patients from preventable harm, strengthen trust, and uphold professional integrity.

Quality Improvement

- Apply quality improvement methods (e.g., audit, evaluation) to monitor and enhance care.
- Analyse and interpret patient, staff, and system data to inform service improvements.

Patient Safety and Incident Management

- Apply safe practices in prescribing, procedures, referrals, infection prevention and control, care transitions, and near-patient diagnostics.
- Identify, escalate, and report risks, incidents, near-misses, and notifiable events in line with statutory and professional duties.
- Participate in open disclosure after adverse events, in line with statutory duty.

Infection Prevention and Control

- Implement evidence-based infection prevention and control, including hand hygiene, aseptic technique, safe PPE use, and safe management of medical devices and clinical environments.

System Safety and Governance

- Demonstrate understanding of local governance structures, reporting systems, and escalation pathways.
- Recognise and escalate organisational or service barriers (e.g., unsafe premises, processes, or systems) that compromise patient safety or timely access to care.

Antimicrobial Resistance

- Understand behavioural, social, environmental, and geographic drivers of antimicrobial resistance in clinical decision-making.

2 Relating to Patients

Doctors must foster respectful, person-centred clinical relationships that uphold patient autonomy, dignity, and trust. This requires clear communication, protection of confidentiality, and supporting informed consent, while recognising individual needs and potential barriers to care. By practising with fairness and shared responsibility, doctors enable patients to participate meaningfully in decisions about their care and contribute to safer, more equitable outcomes.

Person-Centred Care

- Deliver care that upholds dignity, autonomy, and individual preferences, considering cultural context and social determinants of health.
- Communicate clearly and accessibly, adapting to patients' language, literacy, cognitive ability, and circumstances.

Confidentiality

- Protect confidentiality across all communications, applying data-protection legislation and managing required disclosures appropriately.
- Explain limits to confidentiality where required (e.g. safeguarding, public health, or legal duties).

Informed Consent and Shared Decision-Making

- Assess capacity and ensure discussions allow sufficient time to explain risks, benefits, and alternatives, and, where relevant, the purpose and implications of complex or sensitive procedures (e.g., genetic testing).
- Support patient autonomy through informed consent and shared decision-making, respecting valid Advance Healthcare Directives when capacity is lacking.
- Identify, address or escalate cultural and social barriers to participation in healthcare decisions.

Information and Care Navigation

- Provide clear, balanced, and evidence-based information to help patients understand their care options, make informed decisions, and access appropriate services or supports.
- Coordinate referrals and share relevant information to support continuity and navigation of care pathways.

Relationships and Boundaries

- Build respectful relationships with patients while maintaining professional boundaries.
- Be clear about the limits of competence and refer patients when required.

Health Promotion and Preventive Care

- Provide evidence-based health promotion and preventive care advice, tailored to individual risk factors.

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3 Communication and Interpersonal Skills

Doctors must communicate clearly, compassionately, and safely with patients, families, and colleagues to support trust, understanding, and shared decision-making. This requires adapting communication to meet individual needs, handling challenging conversations with sensitivity, upholding professional boundaries, and ensuring accuracy in records, correspondence, and handovers. By communicating effectively across all settings, doctors reduce risk, ensure patient understanding, and promote safe, coordinated care.

Clinical Communication and Documentation

- Take accurate, structured histories and explain diagnoses, care plans, and clinical decisions with clarity and empathy.
- Apply handover protocols to ensure safe care transitions.
- Maintain complete, timely, and legible documentation to support continuity, safety, and compliance

Patient Communication and Comprehension

- Confirm and document patient understanding of information shared, including risks, benefits, alternatives, and limitations.
- Apply health literacy principles across verbal, written, digital, and visual formats.
- Deliver difficult news clearly and with empathy.
- Adapt communication to patient capacity, language, literacy, cognitive ability, or culture, involving interpreters, advocates, or supports (e.g., written materials) as required.

Safeguarding

- Conduct safeguarding discussions respectfully, protecting dignity, confidentiality, and legal compliance.
- Escalate safeguarding concerns through appropriate channels.

Complaints and Regulatory Communication

- Respond promptly and professionally to patient complaints and enquiries.
- Reduce complaint risk through clear communication, accurate records, and timely follow-up.
- Engage constructively with organisational and regulatory complaint processes and contribute to service learning.

Open Disclosure

- Participate in supervised open disclosure discussions after adverse events using honest, transparent, and compassionate communication, in line with statutory and professional standards.

Team Dialogue

- Engage in respectful and constructive dialogue with colleagues to support shared understanding and safe decisions.
- Identify and escalate communication breakdowns that may compromise patient safety.

4 Collaboration and Teamwork

Doctors must work collaboratively with colleagues across disciplines and services to deliver safe, coordinated, and high-quality care. This requires contributing to shared decisions, respecting team roles, and maintaining open and constructive communication. By promoting collaboration and teamwork, doctors strengthen service delivery, promote shared accountability, and foster continuous improvement in team-based care.

Governance and Organisational Awareness

- Understand local governance and leadership structures relevant to your role, including responsibilities and lines of accountability.
- Raise clinical, safety, resource, or organisational concerns through appropriate channels in line with governance and escalation policies

Team Coordination and Integrated Care

- Build effective working relationships with interprofessional teams, recognising the roles of all members.
- Share accountability for decision-making and care coordination, recognising the risks of fragmentation.
- Ensure continuity of care by providing timely, accurate discharge summaries.

Organisational Leadership and Team Culture

- Contribute to leadership by facilitating shared decision-making, coordinating care, and supporting junior colleagues.
- Foster psychological safety by promoting respectful communication, shared learning, and open dialogue.
- Manage conflict to support respectful, functional, and safe team environments.

Team Learning and Development

- Engage in structured team-based learning (e.g., case reviews, safety forums), to inform service improvement and professional development.
- Provide and receive feedback constructively to support team development and patient care quality.

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5 Management (Including Self-Management)

Doctors must manage workload, time, and personal wellbeing to ensure safe and effective clinical practice. This requires prioritising tasks, recognising limits, escalating concerns appropriately, and engaging constructively with organisational systems and processes. By balancing personal capacity with service demands, doctors protect patients from harm, prevent burnout, and support the safe and sustainable delivery of healthcare.

Health, Wellbeing, and Development

- Monitor personal health and performance, recognising fatigue or burnout, and seek support when needed.
- Set and review professional development goals informed by reflection, supervision, and feedback.

Workload and Task Management

- Prioritise tasks to deliver timely, safe, and effective care.
- Coordinate rotas, leave, handovers, and cover to maintain service continuity.
- Communicate availability and scheduling clearly to colleagues.

Administrative Competence

- Complete documentation and administrative tasks accurately and on time.
- Engage with training and professional development, including preparation, participation, and submission of required materials.
- Fulfil supervisory and/or line-management responsibilities where appropriate (e.g., supporting colleagues, approving leave, and contributing to performance assessments).
- Use operational tools (e.g., rotas, workflows, IT systems) effectively to support safe and coordinated care.

Sustainability and Environmental Stewardship

- Order, prescribe, investigate, and deliver care responsibly, ensuring clinical necessity while adopting resource-conscious and sustainable approaches.
- Be aware of organisational sustainability initiatives (e.g., green prescribing, waste reduction).

Systems and Safety Engagement

- Recognise how system pressures (e.g. staffing levels) affect patient safety.
- Contribute to local safety monitoring, governance, and service improvement activities within role and training scope.

6 Scholarship

Doctors should maintain and advance their professional competence through lifelong learning, supervision, reflection, teaching, and research. This requires engaging critically with evidence, translating learning into practice improvement, and contributing to the education and development of colleagues. By integrating inquiry, reflection, and shared learning into their work, doctors strengthen decision-making, enhance patient safety, and uphold professional standards.

Evidence-Based Practice

- Apply research evidence, guidelines, and clinical data appropriately to inform patient care.
- Use audit, service evaluation, and quality improvement data to evaluate and improve practice.

Lifelong Learning and Scope of Practice

- Comply with training and development requirements within your training programme (e.g., maintaining your ePortfolio).
- Set and evaluate learning goals informed by reflection, feedback, and supervision.
- Use insights from audits, reviews, and adverse events to improve practice.
- Recognise limits in knowledge or skill and seek supervision or escalate when required.

Teaching and Role Modelling

- Teach, supervise, and support colleagues and teams using effective communication and evidence-based practice.
- Share clinical knowledge to strengthen team learning and service improvement.
- Model professionalism, clinical integrity, critical thinking and reflective practice in everyday work.

Research and Dissemination

- Undertake audit, research, or service evaluation, disseminating and communicating findings through professional or academic channels.
- Comply with legal, institutional, and ethical standards in research activities.

Innovation and Digital Literacy

- Apply health informatics, telehealth, and emerging technologies with attention to safety, evidence base, and ethical considerations.
- Evaluate risks, benefits, and limitations of digital innovations, including AI, to ensure safe and effective patient care.

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7 Professionalism

Doctors must uphold integrity, accountability, and respect in all aspects of clinical care, leadership, and professional practice. This requires complying with legal and regulatory duties, maintaining confidentiality and professional boundaries, and acting with fairness in healthcare delivery. By modelling professionalism, doctors build trust, protect patients, and promote safe, inclusive healthcare systems.

Statutory and Ethical Duties

- Comply with legal and regulatory requirements, reporting unsafe or unprofessional behaviours, and engaging with investigations and complaints.
- Fulfil safeguarding duties, including mandatory reporting of child protection and vulnerable adult concerns.
- Uphold professional boundaries across all settings to protect patient dignity, autonomy, and trust.
- Protect patient data in line with GDPR and professional standards.
- Declare and transparently manage conflicts of interest in clinical, research, and public activities.

Resource Use and Stewardship

- Use diagnostic, prescribing, and other clinical resources responsibly and fairly, ensuring clinical justification.
- Integrate sustainability principles into practice, balancing immediate patient needs with long-term system and environmental responsibility.

Advocacy, Equity and Fair Practice

- Treat patients and colleagues with dignity and respect, ensuring care is free from discrimination.
- Advocate for fair access to, and equitable experience within, healthcare by recognising and addressing diverse needs and social or structural barriers, inclusive of disability and socioeconomic disadvantage.

Antimicrobial Stewardship

- Prescribe antimicrobials responsibly, selecting agents, dosing, and duration appropriately.
- Participate in stewardship initiatives, such as audits, surveillance, and outbreak management.

Professional Leadership and Accountability

- Represent the profession with integrity, modelling leadership that promotes a culture of safety, openness, and professional accountability.
- Take responsibility for patient safety by identifying and escalating risks, and contributing to learning (e.g., AAR, NIMS).
- Manage personal or team workload pressures, escalating where necessary to maintain safe practice.
- Recognise and respond to signs of stress or impaired performance in self and colleagues, addressing appropriately to safeguard wellbeing and team function.

Public and Online Professional Conduct

- Uphold professional standards in all online and social media activity, recognising that the same expectations apply as in face-to-face communication.
- Maintain patient confidentiality and clear boundaries, separating personal and professional use, and directing patient contact through formal channels.
- Ensure that public communications are accurate, evidence-based, and compliant with regulatory standards.

8 Clinical Skills

Doctors must maintain and apply clinical skills that enable safe, accurate, and effective assessment, diagnosis, and treatment across all stages of patient care. This requires integrating patient history, examination findings, investigations, and patient context to inform clinical reasoning, safe prescribing, and appropriate escalation or referral. By applying these skills responsibly, doctors support patient safety, ensure continuity of care, and deliver high-quality outcomes across healthcare settings. This Domain addresses the professional and ethical responsibilities that underpin the safe application of practice, complementing specialty-specific technical competencies.

Assessment and Reasoning

- Conduct comprehensive assessments, with consent, integrating history, examination, investigations, and patient context.
- Apply structured reasoning to generate differential diagnoses and safe management plans, using evidence and guidelines.
- Recognise uncertainty, limits of competence, or impaired performance, and escalate or seek supervision when required.
- Take account of the patient's psychological, social, and contextual factors where clinically relevant to safe decision-making.
- Use digital tools responsibly to support assessment, decision-making, and care delivery.

Transfer of Care

- Refer or transfer patients as required, contributing to collaboration, coordination, and continuity across services.

Records and Communication

- Maintain accurate and timely records and correspondence to support safe handover, discharge, and care transitions, complying with legal and data protection standards.

Complex Care Planning

- Initiate and participate in discussions regarding high-risk or complex care, including end-of-life care and advanced planning, ensuring shared decision-making.
- Provide person-centred care for patients with life-limiting illness, including pain and symptom control, and family support.

Safe Prescribing

- Prescribe safely and appropriately, selecting the correct drug, dose, route, and duration, and ensure monitoring or handover where required.

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SPECIALTY SECTION – CLINICAL GENETICS TRAINING GOALS

This section includes the Clinical Genetics Training Goals that the Trainee should achieve by the end of Higher Specialist Training.

Each Training Goal is broken down into specific and measurable Training Outcomes.

Under each Outcome there is an indication of the suitable and recommended training/learning opportunities and assessment methods.

In order to achieve the Outcomes it is recommended to agree on the most appropriate type of training and assessment methods with the assigned Trainer.

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Training Goal 1 – Manage a Comprehensive Clinical & Biochemical Genetic Medicine Service for Fetal, Paediatric, & Adult Patients

By the end of HST Trainees will be able to manage a comprehensive clinical and biochemical genetic medicine service for fetal, paediatric, and adult patients.

Outcome 1 – Demonstrate understanding of genetics, cellular, and molecular mechanisms underpinning clinical biochemical and biochemical genetics

Training/Learning Opportunities

Self-directed learning
Study Days
Course Attendance
Feedback opportunities
WBA

Outcome 2 – Conduct essential pre-clinic preparation, including obtaining prior medical reports and undertaking appropriate literature review

Training/Learning Opportunities

Clinic Attendance
Consultations
Feedback opportunities
WBA

Outcome 3 – Take, document, and interpret a detailed personal, and family history

Training/Learning Opportunities

Clinic Attendance
Feedback opportunities
WBA

Outcome 4 – Undertake and document clinical examination relevant to the condition

Training/Learning Opportunities

Clinical Attendance
Consultations
Feedback opportunities
WBA

Outcome 5 – Formulate differential diagnoses including undertaking genotype/phenotype correlation where appropriate

Training/Learning Opportunities

Clinic Attendance
Literature review
Attendance at appropriate course(s)
Feedback opportunities

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Outcome 6 – Discuss, document, and arrange appropriate testing

Training/Learning Opportunities

Clinic attendance
Feedback opportunities
WBA

Outcome 7 – Obtain and document informed consent including consent in special circumstances and testing limitations and sample storage

Training/Learning Opportunities

Clinic Attendance
Feedback opportunities
WBA

Outcome 8 – Use principles of non-directive counselling

Training/Learning Opportunities

Clinic Attendance
Feedback opportunities
WBA

Outcome 9 – Explain the genetic basis of family condition to affected or at risk individuals

Training/Learning Opportunities

Clinic Attendance
Course Attendance (Communications)
Feedback opportunities
WBA

Outcome 10 – Interpret, report, and explain genetic test results and the implications for the individual and the family

Training/Learning Opportunities

Clinic attendance
Consultations
Feedback opportunities
WBA

Outcome 11 – Write appropriate letters to patients and clinicians summarising the diagnosis risk assessment, reproductive options and/or medical recommendations and family implications where appropriate

Training/Learning Opportunities

Consultations
Feedback opportunities
WBA

Outcome 12 – Signpost patients/clinicians to appropriate resources

Training/Learning Opportunities
Consultations
Feedback opportunities
WBA

Training Goal 2 – Work Within Multidisciplinary Teams

By the end of HST Trainees are expected to be able to work within clinical and laboratory multidisciplinary teams (local/national/international) for the management, treatment, and prevention of genetic disorders.

Outcome 1 – Participate (observe and contribute) in multidisciplinary meetings and clinics

Training/Learning Opportunities

Attend MDT
Consultations
Feedback opportunities
WBA

Outcome 2 – Be aware of the appropriate European reference networks (e.g., Ithaca, METAB ERN) and participate in European meetings (e.g., European Reference Networks) if appropriate

Training/Learning Opportunities

Attend appropriate conferences
Feedback opportunities
WBA

Outcome 3 – Organise and lead local MDT's and implement MDT decisions

Training/Learning Opportunities

Attend/Lead MDTs
Feedback opportunities
WBA

Outcome 4 – Prioritise therapeutic interventions

Training/Learning Opportunities

Clinic Attendance
Conference Attendance
Feedback opportunities
WBA

Outcome 5 – Work closely with genetic counsellors and laboratory staff to optimise patient and family management

Training/Learning Opportunities

Liaison with other specialties/healthcare professionals/laboratory
Laboratory time
Feedback opportunities
WB

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Training Goal 3 – Manage Genetic, Genomic, & Biochemical Testing

By the end of HST Trainees will be able to understand and/or manage genetic, genomic, and biochemical testing on an individual, familial, and population level

OUTCOME 1 – EXPLAIN THE IMPLICATIONS AND LIMITATIONS OF DIAGNOSTIC, PREDICTIVE, AND CASCADE TESTING AND IMPLEMENT APPROPRIATELY

Training/Learning Opportunities

Clinic Attendance
Consultations
Feedback opportunities
WBA

OUTCOME 2 – CRITICALLY APPRAISE THE CLINICAL AND ANALYTICAL VALIDITY AND UTILITY OF TESTS

Training/Learning Opportunities

Time spent in laboratory
Self-directed learning
Feedback opportunities
WBA

OUTCOME 3 – CHOOSE THE APPROPRIATE TEST BASED ON PATIENT CIRCUMSTANCES, CLINICAL VALIDITY AND UTILITY, ANALYTICAL VALIDITY, AND ETHICAL CONSIDERATIONS

Training/Learning Opportunities

Clinic Attendance
Time spent in laboratory
Self-directed learning
Feedback opportunities
WBA

OUTCOME 4 – EXPLAIN THE IMPLICATIONS OF INCIDENTAL AND SECONDARY GENETIC FINDINGS

Training/Learning Opportunities

Clinic Attendance
Feedback opportunities
WBA

OUTCOME 5 – DEMONSTRATE AWARENESS OF POPULATION GENE FREQUENCIES AND ITS EFFECT ON TESTING STRATEGIES

Training/Learning Opportunities

Attendance at appropriate courses
Feedback opportunities
WB

Training Goal 4 – Manage Reproductive & Perinatal Genetic Cases

By the end of HST Trainees are expected to be able to manage reproductive and perinatal genetic cases and other post-mortem genetic testing

Outcome 1 – Discuss appropriate reproductive options in a non-directive manner with patients

Training/Learning Opportunities

Clinic Attendance
Course Attendance
Feedback opportunities
WBA

Outcome 2 – Explain and interpret genetic testing in the pre-and peri-natal setting including pre-natal diagnosis, pre-implantation genetic diagnosis and non-invasive pre-natal diagnosis when applicable

Training/Learning Opportunities

Clinic Attendance
Consultations
Feedback opportunities
WBA

Outcome 3 – Awareness of the legal framework underpinning reproductive, obstetric, and fetal medicine practice

Training/Learning Opportunities

Attendance at relevant courses
Self-directed learning
Feedback opportunities
WBA

Outcome 4 – Manage the appropriate handling of genetic data or material after the death of a patient with a suspected genetic condition

Training/Learning Opportunities

Attendance at appropriate courses
Feedback opportunities
WB

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Training Goal 5 – Evaluate & Interpret Genomic, Biochemical & Clinical Data

By the end of HST Trainees will be able to evaluate and interpret genomic, biochemical, and clinical data and understand the scientific basis underpinning and linking them

OUTCOME 1 – DEMONSTRATE UNDERSTANDING AND APPLICATION OF LABORATORY TECHNIQUES THAT UNDERPIN CURRENT GENETIC TESTING INCLUDING IMPORTANCE OF SAMPLE TYPE AND ORIGIN

Training/Learning Opportunities

Laboratory liaison
Course Attendance
Feedback opportunities
WBA

OUTCOME 2 – DESCRIBE THE PRINCIPLES GOVERNING VARIANT INTERPRETATION AND APPLY THE PRINCIPLES TO ASSESS THE PATHOGENICITY OF A SPECIFIC VARIANT INCLUDING THE USE OF APPROPRIATE RESOURCES (E.G., GENOMIC DATABASES)

Training/Learning Opportunities

Self-directed learning
Attendance at study days
Course Attendance
Feedback opportunities
WBA

OUTCOME 3 – CRITICALLY APPRAISE RESULTS INCLUDING RELEVANCE TO PHENOTYPE AND ASSESS UTILITY TO PATIENT CARE

Training/Learning Opportunities

Self-directed learning
Clinic Attendance
Laboratory Liaison
Attendance at study days
Course Attendance
Feedback opportunities
WBA

OUTCOME 4 – RECOGNISE THAT CLASSIFICATION OF VARIANT PATHOGENICITY CAN CHANGE OVER TIME AND UNDERSTAND THE IMPORTANCE OF REVISITING RESULTS

Training/Learning Opportunities

Self-directed learning
Laboratory Liaison
Attendance at study days
Course Attendance
Feedback opportunities
WBA

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OUTCOME 5 – UNDERSTANDING OF LABORATORY ACCREDITATION AND LABORATORY QUALITY MANAGEMENT SYSTEMS

Training/Learning Opportunities

Self-directed learning
Laboratory Liaison
Attendance at study days
Course Attendance
Feedback opportunities
WBA

Training Goal 6 – Apply Principles of Research, Ethics, & Data Management

By the end of HST Trainees are expected to demonstrate understanding and application of research, ethics, data management to genetic practice

Outcome 1 – Recognise importance of research in patient care and critically appraise scientific literature

Training/Learning Opportunities

Research work
Presentations
Audit
Feedback opportunities
WBA

Outcome 2 – Recognise the importance of clinical trials, how to source clinical trials and signpost trial opportunities to patients

Training/Learning Opportunities

Participate in clinical trial
Research work
Self-directed learning
Feedback opportunities
WBA

Outcome 3 – Demonstrate contribution to research (e.g., publications, presentations, enrolling patients in research)

Training/Learning Opportunities

Feedback opportunities
WBA
Presentations
Publications

Outcome 4 – Recognise ethical issues specific to clinical genetics (e.g., familial nature of genetic information, ethical impact of new genetic technologies including gene therapy and newborn screening)

Training/Learning Opportunities

Course Attendance
Clinic attendance
Participation in laboratory meetings
Feedback opportunities
WBA

Outcome 5 – Recognise data management issues specific to clinical genetics (e.g., sharing of familial genetic data, issues of security and anonymisation of genomic data)

Training/Learning Opportunities

Attendance at appropriate courses
Clinic attendance
Feedback opportunities
WBA

APPENDICES

This section includes two appendices to the Curriculum.

The first one is about Assessment (i.e. Workplace Based Assessments, Evaluations etc).

The second one is about Teaching Attendance (i.e. Taught Programme, Specialty-Specific Learning Activities and Study Days)

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ASSESSMENT APPENDIX

Workplace-Based Assessment & Evaluations

The expression "workplace-based assessments" (WBA) defines all the assessments used to evaluate Trainees' daily clinical practices employed in their work setting. It is primarily based on the observation of Trainees' performance by Trainers. Each observation is followed by a Trainer's feedback, with the intent of fostering reflective practice.

Relevance of Feedback for WBA

Although "assessment" is the keyword in WBA, it is necessary to acknowledge that feedback is an integral part and complementary component of WBA. The main purpose of WBA is to provide specific feedback for Trainees. Such feedback is expected to be:

- **Frequent**: the opportunities to provide feedback are preferably given by directly observed practice, but also by indirectly observed activities. Feedback is expected to be frequent and should concern a low-stake event. Rather than being an assessor, the Trainer is an observer who is asked to provide feedback in the context of the training opportunity presented at that moment.
- **Timely**: preferably, the feedback should be a direct conversation between Trainer and Trainee in a timeframe close to the training event. The Trainee should then record the feedback on ePortfolio in a timely manner.
- **Constructive**: the recorded feedback would inform both Trainee's practice for future performance and committees for evaluations. Hence, feedback should provide Trainees with behavioural guidance on how to improve performance and give committees the context that leads to a rating, so that progression or remediation decisions can be made.
- **Actionable**: to improve performance and foster behavioural change, feedback should include practical and contextualised examples of both Trainee's strengths and areas for improvement. Based on these examples, it is necessary to outline a realistic action plan to direct the Trainee towards remediation/improvement.

Types of WBAs in use at RCPI

There is a variety of WBAs used in medical education. They can be categorised into three main groups: *Observation of performance*; *Discussion of clinical cases*; *Feedback*; *Mandatory Evaluations*.

As WBAs at RCPI we use *Observation of performance* via MiniCEX and DOPS; *Discussion of clinical cases* via CBD; *Feedback* via Feedback Opportunity.

Mandatory Evaluations are bound to specific events or times of the academic year, for these at RCPI we use: Quarterly Assessment/End of Post Assessment; End of Year Evaluation; Penultimate Year Evaluation; Final Year Evaluation.

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Recording WBAs on ePortfolio

It is expected that WBAs are logged on an electronic portfolio. Every Trainee has access to an individual ePortfolio where they must record all their assessments, including WBAs. By recording assessments on this platform, ePortfolio serves both the function to provide an individual record of the assessments and to track Trainees' progression.

Formative & Summative Assessment

The Trainee can record any WBA either as formative or summative with the exception of the *Mandatory Evaluations* (Quarterly/End of Post, End of Year, Penultimate Year, Final Year evaluations).

If the WBA is logged as formative, the Trainee can retain the feedback on record, but this will not be visible to an assessment panel, and it will not count towards progression. If the WBA is logged as summative it will be regularly recorded and it will be fully visible to assessment panels, counting towards progression.

Specialty-Specific Examination

Sit the European Board of Medical Genetics **or** Royal College of Pathologists Certificate in Medical Genetics These exams will not be used as a certifying or qualifying examinations but are to be used as a self- assessment tool designed to gauge knowledge in Clinical Genetics.

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WORKPLACE-BASED ASSESSMENTS	
CBD *|Case Based Discussion*	This assessment is developed in three phases: 1. Planning: The Trainee selects two or more medical records to present to the Trainer who will choose one for the assessment. Trainee and Trainer identify one or more training goals in the curriculum and specific outcomes related to the case. Then the Trainer prepares the questions for discussion. 2. Discussion: Prevalently, based on the chosen case, the Trainer verifies the Trainee's clinical reasoning and professional judgment, determining the Trainee's diagnostic, decision-making and management skills. 3. Feedback: The Trainer provides constructive feedback to the Trainee. It is good practice to complete at least one CBD per quarter in each year of training.
DOPS | *Direct Observation of Procedural Skills*	This assessment is specifically targeted at the evaluation of procedural skills involving patients in a single encounter. In the context of a DOPS, the Trainer evaluates the Trainee while they are performing a procedure as a part of their clinical routine. This evaluation is assessed by completing a form with pre-set criteria, then followed by direct feedback.
MiniCEX | *Mini Clinical Examination Exercise*	The Trainer is required to observe and assess the interaction between the Trainee and a patient. This assessment is developed in three phases: 1. The Trainee is expected to conduct a history taking and/or a physical examination of the patient within a standard timeframe (15 minutes). 2. The Trainee is then expected to suggest a diagnosis and management plan for the patient based on the history/examination. 3. The Trainer assesses the overall Trainee's performance by using the structured ePortfolio form and provides constructive feedback.
Feedback Opportunity	Designed to record as much feedback as possible. It is based on observation of the Trainees in any clinical and/or non-clinical task. Feedback can be provided by anyone observing the Trainee (peer, other supervisors, healthcare staff, juniors). It is possible to turn the feedback into an assessment (CDB, DOPS or MiniCEX)
MANDATORY EVALUATIONS	
QA | *Quarterly Assessment*	As the name suggests, the Quarterly Assessment recurs four times in the academic year, once every academic quarter (every three months). It frequently happens that a Quarterly Assessment coincides with the end of a post, in which case the Quarterly Assessment will be substituted by completing an End of Post Assessment. In this sense the two Assessments are interchangeable, and they can be completed using the same form on ePortfolio. However, if the Trainee will remain in the same post at the end of the quarter, it will be necessary to complete a Quarterly Assessment. Similarly, if the end of a post does not coincide with the end of a quarter, it will be necessary to complete an End of Post Assessment to assess the end of a post. This means that for every specialty and level of training, a minimum of four Quarterly Assessment and/or End of Post Assessment will be completed in an academic year as a mandatory requirement.
EOPA | *End of Post Assessment*	
EOYE | *End of Year Evaluation*	The End of Year Evaluation occurs once a year and involves the attendance of an evaluation panel composed of the National Specialty Directors (NSDs); the Specialty Coordinator attends too, to keep records of and facilitate the meeting. The assigned Trainer is not supposed to attend this meeting unless there is a valid reason to do so. These meetings are scheduled by the respective Specialty Coordinators and happen sometime before the end of the academic year (between April and June).
PYE | *Penultimate Year Evaluation*	The Penultimate Year Evaluation occurs in place of the End of Year Evaluation, in the year before the last year of training. It involves the attendance of an evaluation panel composed of the National Specialty Directors (NSDs) and an External Member who is a recognised expert in the Specialty outside of Ireland; the Specialty Coordinator attends too, to keep records of and facilitate the meeting. The assigned Trainer is not supposed to attend this meeting unless there is a valid reason to do so.
FYE | *Final Year Evaluation*	In the last year of training, the End of Year Evaluation is conventionally called Final Year Evaluation, however, its organisation is the same as an End of Year Evaluation.

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TEACHING APPENDIX

RCPI Taught Programme

The RCPI Taught Programme consists of a series of modular elements spread across the years of training.

Delivery will be a combination of self-paced online material, live virtual tutorials, and in-person workshops, all accessible in one area on the RCPI's virtual learning environment (VLE), RCPI Brightspace.

The live virtual tutorials will be delivered by Tutors related to this specialty and they will use specialty-specific examples throughout each tutorial. Trainees will be assigned to a tutorial group and will remain with their tutorial group for the duration of HST.

Trainees will receive their induction content and timetable ahead of their start date on HST. Trainees must plan the time to complete their requirements and must be supported with the allocation of study leave or appropriate rostering.

As the HST Taught Programme is a mandatory component of HST, it is important that Trainees are released from service to attend the Virtual Tutorials and, where possible facilitated with the use of teaching space in the hospital.

Specialty-Specific Learning Activities (Courses & Workshops)

Trainees will also complete specialty-specific courses and/or workshops as part of the programme.

Trainees should always refer to their training curriculum for a full list of requirements for their HST programme. When not sure, Trainees should contact their Programme Coordinator.

Study Days

Study days vary from year to year, they comprise a rolling schedule of hospital-provided topic-specific educational days and national/international events selected for their relevance to the HST curriculum.

Trainees are expected to attend the majority of the study days available and **at least 2 per training year**.

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Clinical Genetics Teaching Attendance Requirements

RCPI Taught Programme

Modularised content available on Brightspace

Specialty-Specific Learning Activities

Specialty Courses

- European Recognised Online or Face to Face Clinical Bioinformatics Course (tp ne agreed with Trainer, examples - Wellcome Trust Sanger Centre Cambridge Bioinformatics Course
- Educational Material about NGS and genome seqiuencing on US Clingen site
- ERN/ESHG - sites and webinars
- ESHG Clinical Genomics and NGS Course, Bertinoro
- European recognised online or face to face genetic counselling course (ato be agreed with Trainer)
- ESHG supported genetics and genomics courses (desirable)
- Genomic Training Academy/Genomic Training Programme
- RCPI Cancer Genetics (Desirable)

Study Days

Trainees are expected to attend the majority of the study days available and

at least 2 per training year

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