INSTITUTE
OF MEDICINE

ROYAL COLLEGE OF
PHYSICIANS OF IRELAND

HIGHER SPECIALIST TRAINING IN

Clinical Genetics

OUTCOME-BASED EDUCATION – OBE CURRICULUM

This Curriculum of Higher Specialist Training in Clinical Genetics was developed in 2023 by a working group led by Professor Andrew Green, National Specialty Director, and the RCPI Education Department. 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
1.0	01 July 2024	Keith Farrington	New OBE Curriculum

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.

Table of Contents

1. INTRODUCTION

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

1.1. 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.

1.2. 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.

1.3. 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.

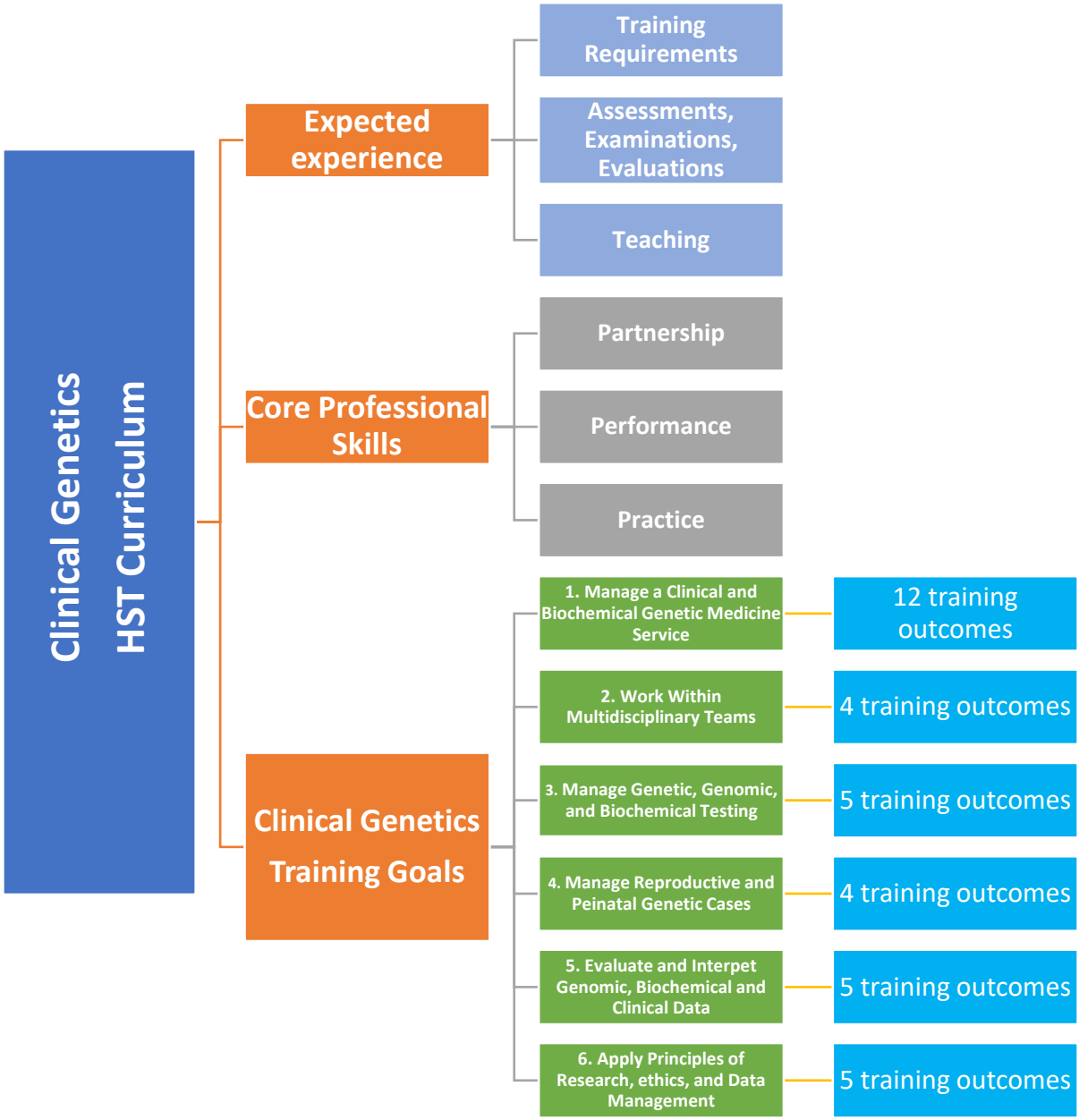
It is important to note that ePortfolio is a digital repository designed to reflect Curriculum requirements. It facilitates recording of progress through HST and evidence that training is valid and appropriate. While a complete ePortfolio is essential for HST certification, Trainees and Trainers should 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.

1.4. Reference to rules and 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.

1.5. Overview of Curriculum Sections and Training Goals



2. EXPECTED EXPERIENCE

This section details the training experience and the service provision tasks that all Trainees are expected to complete throughout the Higher Specialist Training.

2.1. 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.

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.

2.2. Outpatient Clinics, Ward Rounds, Consultations, and 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	
Cardiac Genetics	Years 1-4	20 per programme (in appropriate post)	
Adult Neurogenetics	Years 1-4	20 per programme (in appropriate post)	
Ophthalmology Genetics	Years 1-4	10 per programme (in appropriate post)	
Cystic Fibrosis	Years 1-4	Attend where appropriate	
Cancer	Years 1-4	Attend at least 1 per week in appropriate post	
Neurofibromatosis/Tuberous sclerosis	Years 1-4	Attend at least 1 per week	
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	Years 1-4		
• Medicine (Cardiology)	Years 1-4	Monthly – to attend in appropriate post	
• Paediatric cancer genetics (Crumlin)	Years 1-4	Monthly – to attend in appropriate post	

• Cross City (Paediatrics) Neurology meeting	Years 1-4	Twice yearly – to attend in appropriate post	
• Paediatric Disorders of Sexual Development (Endocrine)	Years 1-4	Twice yearly – to attend in appropriate post	
• Dublin tri-hospital Fetal Medicine group	Years 1-4	Thrice yearly – to attend in appropriate post	
• Variant interpretation meetings	Years 1-4	Every 2 months – to attend in appropriate post	
Procedures	Years 1-4		
• Skin biopsy		As required	
• Buccal Swab		As required	
Laboratory Experience	Years 1-4	As required	
Examinations (either of)	Years 1-4		Examinations
European Board of Medical Genetics	Years 1-4	1 Per Programme	
Royal College of Pathologists Certificate in Medical Genetics	Years 1-4	1 Per Programme	

2.3. Hospital Based Learning and 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**

2.4. Research, Audit, and 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

2.5. 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.

2.6. Assessments, Evaluations, and 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.

2.7. 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

3. CORE PROFESSIONAL SKILLS

This section includes the Medical Council guidelines for medical professional conduct, regarding Partnership, Performance and Practice

Partnership

Communication and interpersonal skills

- Facilitate the exchange of information, be considerate of the interpersonal and group dynamics, and have a respectful and honest approach
- Engage with patients and colleagues in a respectful manner
- Actively listen to the thoughts, concerns, and opinions of others
- Consider data protection, duty of care and appropriate modes of communication when exchanging information with others

Collaboration

- Collaborate with patients, their families, and your colleagues to work in the best interest of the patient, for improved services and to create a positive working environment
- Work cooperatively with colleagues and team members to deliver an excellent standard of care
- Seek to build trust and mutual respect with patients
- Appropriately share knowledge and information, in compliance with GDPR guidelines
- Take on-board available, relevant feedback

Health Promotion

- Communicate and facilitate discussion around the effect of lifestyle factors on health and promote the ethical practice of evidence-based medicine
- Seek up-to-date evidence on lifestyle factors that:
    - negatively impact health outcomes
    - increase risk of illness
    - positively impact health and decrease risk factors
- Actively promote good health practices with patients individually and collectively

Caring for patients

- Take into consideration patient's individuality, personal preferences, goals, and the need to provide compassionate and dignified care
- Be familiar with
    - Ethical guidelines
    - Local and national clinical care guidelines
- Act in the patient's best interest
- Engage in shared decision-making and discuss consent

Performance

Patient safety and ethical practice

- Put the interest of the patient first in decisions and actions
- React in a timely manner to issues identified that may negatively impact the patient's outcome
- Follow safe working practices that impact patient's safety
- Understand ethical practice and the medical council guidelines
- Support a culture of open disclosure and risk reporting
- Be aware of the risk of abuse, social, physical, financial, and otherwise, to vulnerable persons

Organisational behaviour and leadership

- The activities, personnel and resources that impact the functioning of the team, hospital, and health care system
- Understand and work within management systems
- Know the impacts of resources and necessary management
- Demonstrate proficient self-management

Wellbeing

- Be responsible for own well-being and health and its potential impact on the provision of clinical care and patient outcomes
- Be aware of signs of poor health and well-being
- Be cognisant of the risk to patient safety related to poor health and well-being of self and colleagues
- Manage and sustain your own physical and mental well-being

Practice

Continuing competence and lifelong learning

- Continually seek to learn, improve clinical skills, and understand established and emerging theories in the practice of medicine
- Meet career requirements including those of the medical council, your employer, and your training body
- Be able to identify and optimise teaching opportunities in the workplace and other professional environments
- Develop and deliver teaching using appropriate methods for the environment and target audience

Reflective practice and self-awareness

- Bring awareness to your actions and decisions and engage in critical appraisal of your own work to drive lifelong learning and improve practice
- Pay critical attention to the practical values and theories which inform everyday practice
- Be aware of your own level of practice and your learning needs
- Evaluate and appraise your decisions and actions with consideration as to what you would change in the future
- Seek to role model good professional practice within the health service

Quality assurance and improvement

- Seek opportunities to promote excellence and improvements in clinical care through the audit of practice, active engagement in and the application of clinical research and the dissemination of knowledge at all levels and across teams
- Gain knowledge of quality improvement methodology
- Follow best practices in patient safety
- Conduct ethical and reproducible research

4. Specialty Section - Clinical Genetics Training Goals

This section includes the Clinical Genetics 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 **suggested** training/learning opportunities and assessment methods.*

To achieve the Outcomes, it is recommended to agree the most appropriate training and assessment methods with the assigned Trainer.

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

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

Training Goal 3 – Manage Genetic, Genomic, and 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 and 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

Training Goal 5 – Evaluate and interpret Genomic, Biochemical and 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

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, and 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

5. APPENDICES

This section includes two appendices to the curriculum.

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

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

ASSESSMENT APPENDIX

Workplace-Based Assessment and 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.

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 and 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.

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.

TEACHING APPENDIX

RCPI Taught Programme

The new 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**.

Clinical Genetics Teaching Attendance Requirements