INSTITUTE
OF MEDICINE

ROYAL COLLEGE OF
PHYSICIANS OF IRELAND

HIGHER SPECIALIST TRAINING IN

CLINICAL GENETICS

This curriculum of training in Clinical Genetics was developed in 2012 and undergoes an annual review by Prof Andrew Green, National Specialty Directors, Dr Ann O'Shaughnessy, Head of Education, and by the Clinical Genetics Training Committee. The curriculum is approved by the Institute of Medicine.

Version	Date Published	Last Edited By	Version Comments
11.0	01 July 2022	Keith Farrington	Changes to form names

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Introduction

Clinical Genetics is the specialty concerned with the diagnosis and management of inherited disorders and birth defects, with the estimation of genetic risks and with genetic counselling of family members. Clinical genetic specialists generally work in multidisciplinary regional genetic centres, in close collaboration with laboratory scientists, clinical co-workers (genetic counsellors) and academic colleagues.

The specialty of Clinical Genetics is constantly changing and the clinical geneticist must take account of new knowledge and molecular developments and alter clinical practice accordingly. S/He will be an information resource for other medical specialists. Clinical geneticists will need a wide range of clinical skills as genetic disorders can affect people of all ages and involve all body systems. Communication skills are particularly important in explaining complex concepts and genetic test results to families enabling them to make informed decisions and choose an appropriate course of action. The majority of patients are seen in an outpatient setting which allows time to explain such concepts to families. Ward referrals are also carried out on request.

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

The core curriculum will provide training in Biochemical Genetics and the principles of multidisciplinary management of well-defined Genetic Disease relevant to Ireland's commitment to provide management as well as education and training in the management of rare disorders (80% of genetic origin) (EC Recommendation on an action in the field of Rare Disease, June 2009).Clinical Biochemical Genetics is that branch of medicine concerned with the study of inborn errors of metabolism, the diagnosis, genetic counselling and management of individuals of all ages with inherited metabolic diseases (IMDs). These conditions have a particular importance in our population due to their high prevalence within the Republic of Ireland and as well-defined examples of multiple and evolving treatment options for genetic (and rare diseases).

Aims

Upon satisfactory completion of specialist training in Clinical Genetics a doctor will be **competent** to undertake comprehensive medical practice in that specialty in a **professional** manner, unsupervised and independently and/or within a team, in keeping with the needs of the healthcare system.

Competencies, at a level consistent with practice in the specialty of Clinical Genetics, will include the following:

- Patient care that is appropriate, effective and compassionate dealing with health problems and health promotion.
- Medical knowledge in the basic biomedical, behavioural and clinical sciences, medical ethics and medical jurisprudence and application of such knowledge in patient care.
- Interpersonal and communication skills that ensure effective information exchange with individual patients and their families and teamwork with other health professionals, the scientific community and the public.
- Appraisal and utilisation of new scientific knowledge to update and continuously improve clinical practice.
- The ability to function as a supervisor, trainer and teacher in relation to colleagues, medical students and other health professionals.
- Capability to be a scholar, contributing to development and research in the field of Clinical Genetics.
- Professionalism.
- Knowledge of public health and health policy issues: awareness and responsiveness in the larger context of the health care system, including e.g. the organisation of health care, partnership with health care providers and managers, the practice of cost-effective health care, health economics and resource allocations.
- Ability to understand health care and identify and carry out system-based improvement of care.

Professionalism

Being a good doctor is more than technical competence. It involves values – putting patients first, safeguarding their interests, being honest, communicating with care and personal attention, and being committed to lifelong learning and continuous improvement. Developing and maintaining values are important; however, it is only through putting values into action that doctors demonstrate the continuing trustworthiness with the public legitimately expect. According to the Medical Council, Good Professional Practice involves the following aspects:

- Effective communication
- Respect for autonomy and shared decision-making
- Maintaining confidentiality
- Honesty, openness and transparency (especially around mistakes, near-misses and errors)
- Raising concerns about patient safety
- Maintaining competence and assuring quality of medical practice

Entry Requirements

Applicants for Higher Specialist Training (HST) in Clinical Genetics must have a certificate of completion in Basic Specialist Training (BST) in either Paediatrics or General Internal Medicine or a specialty of relevance to Clinical or Bio-Chemical Genetics and obtained the MRCPI.

Applicants who have completed BST in *General Internal Medicine, Paediatric Medicine or other relevant specialities who have* appropriate higher clinical examinations in allied specialities, that are deemed equivalent to the MRCPI, may be considered.

Those who do not hold a BST certificate and MRCPI must provide evidence of equivalency.

A period of experience in both General Medicine & Paediatrics at Senior House Officer Grade is considered desirable before entry to HST, although not essential.

Entry on the training programme is at year 1. Deferrals are not allowed on entry to Higher Specialist Training.

Duration of Training and Organisation of Training

Whilst the curriculum is competency-based, the duration of training must meet the European Minimum of 4 years for full time speciality training adjusted accordingly for flexible training.

Core Clinical Genetics Module incorporating Biochemical Genetics
It is essential that the trainee should have a thorough basic training in genetics with emphasis on human aspects. The training should embrace clinical, laboratory and theoretical work. In addition, training should include statistics and an introduction to relevant computer applications. Practical experience is necessary, at a basic level, of cytogenetic, molecular genetics and laboratory biochemical genetics techniques.

Trainees should have an understanding of the investigation, diagnosis and management of inborn errors of metabolism (IMD) and the principles of new-born screening (including knowledge about expanded new-born screening options). Trainees should be competent in the diagnosis of an IMD with familiarity with treatment of management of common conditions such as PKU, organic acidopathies, urea cycle defects, lysosomal disorders and the modalities of treatment (substrate reduction, organ and HSCT/stem cell transplantation, chaperone and substrate inhibitor treatments, enzyme replacement therapy and the principles of gene therapy and gene manipulation therapies.)

Clinical Genetics Research Module
For those trainees following the academic training pathway the following additional objectives need to be achieved.

The majority of trainees will be expected to have undertaken a supervised research project by the end of their training, or alternatively achieved an understanding of the relevant research objectives in the curriculum through alternative means, such as participation in a research study or attending seminars in research methodology.

After entering an approved programme, some trainees wish to spend a longer period in research by stepping aside from clinical training for up to three years. This is dependent on the research being prospectively approved by the College and with the support of the NSD and Dean of Postgraduate Specialist Training. This is acceptable if relevant competencies are achieved; up to one year's credit can be given completion of the programme. Trainees can chose whether or not to include one year of research time towards CSCST and are required to confirm their intention at the time of commencement of the research year.

Trainees must spend the first two years of training in clinical posts in Ireland before undertaking any period of research or Out of Programme Experience (OCPE).

Flexible Training

National Flexible Training Scheme – HSE NDTP

The HSE NDTP operates a National Flexible Training Scheme which allows a small number of Trainees to train part time, for a set period of time.

Overview

- Have a well-founded reason for applying for the scheme e.g. personal family reasons
- Applications may be made up to 12 months in advance of the proposed date of commencement of flexible training and no later than 4 months in advance of the proposed date of commencement
- Part-time training shall meet the same requirements as full-time training, from which it will differ only in the possibility of limited participation in medical activities to a period of at least half of that provided for full-time trainees

Job Sharing - RCPI

The aim of job sharing is to retain doctors within the medical workforce who are unable to continue training on a full-time basis.

Overview

- A training post can be shared by two trainees who are training in the same specialty and are within two years on the training pathway
- Two trainees will share one full-time post with each trainee working 50% of the hours
- Ordinarily it will be for the period of 12 months from July to July each year in line with the training year
- Trainees who wish to continue job sharing after this period of time will be required to re-apply
- Trainees are limited to no more than 2 years of training at less than full-time over the course of their training programme

Post Re-assignment – RCPI

The aim of post re-assignment is to support trainees who have had an unforeseen and significant change in their personal circumstances since the commencement of their current training programme which requires a change to the agreed post/rotation.

Overview:

- Priority will be given to trainees with a significant change in circumstances due to their own disability, it will then be given to trainees with a change in circumstances related to caring or parental responsibilities. Any applications received from trainees with a change involving a committed relationship will be considered afterwards
- If the availability of appropriate vacancies is insufficient to accommodate all requests eligible trainees will be selected on a first come, first serve basis

For further details on all of the above flexible training options, please see the Postgraduate Specialist Training page on the College website www.rcpi.ie

Training Programme

The training programme offered will provide opportunities to fulfil all the requirements of the curriculum of training for the Clinical Genetics programme in accredited training hospitals. Each post within the programme will have a named trainer/educational supervisor and programmes will be under the direction of the National Specialty Director(s) for Clinical Genetics. Programmes will be as flexible as possible consistent with curricular requirements, for example to allow the trainee to develop a sub-specialty interest.

The experience gained through rotation around different departments is recognised as an essential part of HST. It is preferable that a SpR does not remain in the same unit for longer than 2 years of clinical training; or with the same trainer for more than 1 year. However, given that Clinical Genetics is a small speciality, there is flexibility in this respect and a trainee will mostly likely spend 2 years with the same trainer.

Where an essential element of the curriculum is missing from a programme, access to it should be arranged, by day release for example, or if necessary by secondment.

Teaching, Research and Audit

All trainees are required to participate in teaching. They should also receive basic training in research methods, including statistics, so as to be capable of critically evaluating published work.

A period of supervised research relevant to Clinical Genetics is considered highly desirable and will contribute up to 12 months towards the completion of training. Some trainees may wish to spend two or three years in research leading to an MSc, MD, or PhD, by stepping aside from the programme for a time. For those intending to pursue an academic path, an extended period of research may be necessary in order to explore a topic fully or to take up an opportunity of developing the basis of a future career. Such extended research may continue after the CSCST is gained. However, those who wish to engage in clinical medical practice must be aware of the need to maintain their clinical skills during any prolonged period concentrated on a research topic, if the need to re-skill is to be avoided.

Trainees are required to engage in audit during training and to provide evidence of having completed the process.

ePortfolio

The trainee is required to keep their ePortfolio up to date and maintained throughout HST. The ePortfolio will be countersigned as appropriate by the trainers to confirm the satisfactory fulfilment of the required training experience and the acquisition of the competencies set out in the Curriculum. This will remain the property of the trainee and must be produced at the annual Evaluation meeting.

The trainee also has a duty to maximise opportunities to learn, supplementing the training offered with additional self-directed learning in order to fulfil all the educational goals of the curriculum. Trainees must co-operate with other stakeholders in the training process. It is in a SpR's own interest to maintain contact with the Medical Training Department and Dean of Postgraduate Specialist Training, and to respond promptly to all correspondence relating to training. "Failure to co-operate" will be regarded as, in effect, withdrawal from the HST's supervision of training.

At the annual Evaluation, the ePortfolio will be examined. The results of any assessments and reports by educational supervisors, together with other material capable of confirming the trainee's achievements, will be reviewed.

Assessment Process

The methods used to assess progress through training must be valid and reliable. The Curriculum has been re-written, describing the levels of competence which can be recognised. The assessment grade will be awarded on the basis of direct observation in the workplace by consultant supervisors. Time should be set aside for appraisal following the assessment e.g. of clinical presentations, case management, observation of procedures. As progress is being made, the lower levels of competence will be replaced progressively by those that are higher. Where the grade for an item is judged to be deficient for the stage of training, the assessment should be supported by a detailed note which can later be referred to at the Annual Evaluation Meeting. The assessment of training may utilise the Mini-CEX, DOPS and Case Based Discussions (CBD) methods adapted for the purpose. These methods of assessment have been made available by HST for use at the discretion of the NSD and nominated trainer. They are offered as a means of providing the trainee with attested evidence of achievement in certain areas of the Curriculum e.g. competence in procedural skills, or in generic components. Assessment will also be supported by the trainee's portfolio of achievements and performance at relevant meetings, presentations, audit, in tests of knowledge, attendance at courses and educational events.

Annual Evaluation of Progress

Overview

The HST Annual Evaluation of Progress (AEP) is the formal method by which a trainee's progression through her/his training programme is monitored and recorded each year. The evidence to be reviewed by the panel is recorded by the trainee and trainer in the trainee's e-Portfolio.
There is externality in the process with the presence of the National Specialty Director (NSD) and a Chairperson. Trainer's attendance at the Evaluation is mandatory, if it is not possible for the trainer to attend in person, teleconference facilities can be arranged if appropriate. In the event of a penultimate year Evaluation an External Assessor, who is a consultant in the relevant specialty and from outside the Republic of Ireland will be required.

Purpose of Annual Evaluation

- Enhance learning by providing formative Evaluation, enabling trainees to receive immediate feedback, measure their own performance and identify areas for development;
- Drive learning and enhance the training process by making it clear what is required of trainees and motivating them to ensure they receive suitable training and experience;
- Provide robust, summative evidence that trainees are meeting the curriculum standards during the training programme;
- Ensure trainees are acquiring competencies within the domains of Good Medical Practice;
- Assess trainees' actual performance in the workplace;
- Ensure that trainees possess the essential underlying knowledge required for their specialty;
- Inform Medical Training, identifying any requirements for targeted or additional training where necessary and facilitating decisions regarding progression through the training programme;
- Identify trainees who should be advised to consider a change in career direction.

Structure of the Meeting

The AEP panel speaks to the trainee alone in the first instance. The trainee is then asked to leave the room and a discussion with the trainer follows. Once the panel has talked to the trainer, the trainee is called back and given the recommendations of the panel and the outcome of the AEP.
At the end of the Evaluation, all panel members and the Trainee agree to the outcome of the Evaluation and the recommendations for future training. This is recorded on the AEP form, which is then signed electronically by the Medical Training Coordinator on behalf of the panel and trainee. The completed form and recommendations will be available to the trainee and trainers within their ePortfolio.

Outcomes

- Trainees whose progress is satisfactory will be awarded their AEP
- Trainees who are being certified as completing training receive their final AEP
- Trainees who need to provide further documentation or other minor issues, will be given 2 weeks (maximum 8) from the date of their AEP to meet the requirements. Their AEP outcome will be withheld until all requirements have been met.
- Trainees who are experiencing difficulties and/or need to meet specific requirements for that year of training will not be awarded their AEP. A date for an interim AEP will be decided and the trainee must have met all the conditions outlined in order to be awarded their AEP for that year of training. The "Chairperson's Overall Assessment Report" will give a detailed outline of the issues which have led to this decision and this will go the Dean of Postgraduate Specialist Training for further consideration.
- Trainees who fail to progress after an interim Evaluation will not be awarded their AEP.

The Dean of Postgraduate Training holds the final decision on AEP outcomes. Any issues must be brought to the Dean and the Annual Chairperson's Meeting for discussion.

Facilities

A consultant trainer/educational supervisor has been identified for each approved post. He/she will be responsible for ensuring that the educational potential of the post is translated into effective training which is being fully utilized. The training objectives to be secured should be agreed between trainee and trainer at the commencement of each posting in the form of a written training plan. The trainer will be available throughout, as necessary, to supervise the training process.

All training locations approved for HST have been inspected by the medical training department. Each must provide an intellectual environment and a range of clinical and practical facilities sufficient to enable the knowledge, skills, clinical judgement and attitudes essential to the practice of Clinical Genetics to be acquired.

Physical facilities include the provision of sufficient space and opportunities for practical and theoretical study; access to professional literature and information technologies so that self-learning is encouraged and data and current information can be obtained to improve patient management.

Trainees in Clinical Genetics should have access to an educational programme of e.g. lectures, demonstrations, literature reviews, multidisciplinary case conferences, seminars, study days etc., capable of covering the theoretical and scientific background to the specialty. Trainees should be notified in advance of dates so that they can arrange for their release. For each post, at inspection, the availability of an additional limited amount of study leave for any legitimate educational purpose has been confirmed. Applications, supported if necessary by a statement from the consultant trainer, will be processed by the relevant employer.

Generic Components

This chapter covers the generic components which are relevant to HST trainees of all specialties but with varying degrees of relevance and appropriateness, depending on the specialty.
As such, this chapter needs to be viewed as an appropriate guide of the level of knowledge and skills required from all HST trainees with differing application levels in practice.

Good Professional Practice

Objective: Trainees must appreciate that medical professionalism is a core element of being a good doctor and that good medical practice is based on a relationship of trust between the profession and society, in which doctors are expected to meet the highest standards of professional practice and behaviour.

Medical Council Domains of Good Professional Practice: Relating to Patients, Communication and Interpersonal Skills, Professionalism, Patient Safety and Quality of Patient Care.

KNOWLEDGE

Effective Communication

- How to listen to patients and colleagues
- The principles of open disclosure
- Knowledge and understanding of valid consent
- Teamwork
- Continuity of care

Ethics

- Respect for autonomy and shared decision making
- How to enable patients to make their own decisions about their health care
- How to place the patient at the centre of care
- How to protect and properly use sensitive and private patient information in accordance with data protection legislation and how to maintain confidentiality
- The judicious sharing of information with other healthcare professionals where necessary for care following Medical Council Guidelines
- Maintaining competence and assuring quality of medical practice
- How to work within ethical and legal guideline when providing clinical care, carrying research and dealing with end of life issues

Honesty, openness and transparency (mistakes and near misses)

- Preventing and managing near misses and adverse events.
- When and how to report a near miss or adverse event
- Incident reporting; root cause and system analysis
- Understanding and learning from errors
- Understanding and managing clinical risk
- Managing complaints
- Following open disclosure practices
- Knowledge of national policy and National Guidelines on Open Disclosure

Raising concerns about patient safety

- Safe working practice, role of procedures and protocols in optimal practice
- The importance of standardising practice through the use of checklists, and being vigilant
- Safe healthcare systems and provision of a safe working environment
- Awareness of the multiple factors involved in failures
- Knowledge and understanding of Reason's Swiss cheese model
- Understanding how and why systems break down and why errors are made
- Health care errors and system failures
- Human and economic costs in system failures
- The important of informing a person of authority of systems or service structures that may lead to unsafe practices which may put patients, yourself or other colleagues at risk
- Awareness of the Irish Medical Councils policy on raising concerns about safety in the environment in which you work

SKILLS

- Effective communication with patients, families and colleagues
- Co-operation and collaboration with colleagues to achieve safe and effective quality patient care
- Being an effective team player
- Ethical and legal decision making skills
- Minimising errors during invasive procedures by developing and adhering to best-practice guidelines for safe surgery
- Minimising medication errors by practicing safe prescribing principles
- Ability to learn from errors and near misses to prevent future errors
- Managing errors and near-misses
- Using relevant information from complaints, incident reports, litigation and quality improvement reports in order to control risks
- Managing complaints
- Using the Open Disclosure Process Algorithm

ASSESSMENT & LEARNING METHODS

- Consultant feedback at annual assessment
- Workplace based assessment e.g. Mini-CEX, DOPS, CBD
- Educational supervisor's reports on observed performance (in the workplace): prioritisation of patient safety in practice
- RCPI HST Leadership in Clinical Practice
- RCPI Ethics programmes
- Medical Council Guide to Professional Conduct and Ethics
- Reflective learning around ethical dilemmas encountered in clinical practice
- Quality improvement methodology course - recommended

Infection Control

Objective: To be able to appropriately manage infections and risk factors for infection at an institutional level, including the prevention of cross-infections and hospital acquired infection

Medical Council Domains of Good Professional Practice: Patient Safety and Quality of Patient Care; Management (including Self-Management).

KNOWLEDGE

Within a consultation

- The principles of infection control as defined by the HIQA
- How to minimise the risk of cross-infection during a patient encounter by adhering to best practice guidelines available, including the 5 Moments for Hand Hygiene guidelines
- The principles of preventing infection in high risk groups e.g. managing antibiotic use to prevent Clostridium difficile
- Knowledge and understanding of the local antibiotic prescribing policy
- Awareness of infections of concern, e.g. MRSA, Clostridium difficile
- Best practice in isolation precautions
- When and how to notify relevant authorities in the case of notifiable infectious disease
- Understanding the increased risk of infection to patients in surgery or during an invasive procedure and adhering to guidelines for minimising infection in such cases
- The guidelines for needle-stick injury prevention and management

During an outbreak

- Guidelines for minimising infection in the wider community in cases of communicable diseases and how to seek expert opinion or guidance from infection control specialists where necessary
- Hospital policy/seeking guidance from occupational health professional regarding the need to stay off work/restrict duties when experiencing infections the onward transmission of which might impact on the health of others

SKILLS

- Practicing aseptic techniques and hand hygiene
- Following local and national guidelines for infection control and management
- Prescribing antibiotics according to antibiotic guidelines
- Encouraging staff, patients and relatives to observe infection control principles
- Communicating effectively with patients regarding treatment and measures recommended to prevent re-infection or spread
- Collaborating with infection control colleagues to manage more complex or uncommon types of infection including those requiring isolation e.g. transplant cases, immunocompromised host
- In the case of infectious diseases requiring disclosure:
    - Working knowledge of those infections requiring notification
    - Undertaking notification promptly
    - Collaborating with external agencies regarding reporting, investigating and management of notifiable diseases
    - Enlisting / requiring patients' involvement in solving their health problems, providing information and education
    - Utilising and valuing contributions of health education and disease prevention and infection control to health in a community

ASSESSMENT & LEARNING METHODS

- Consultant feedback at annual assessment
- Workplace based assessment e.g. Mini-CEX, DOPS, CBD
- Educational supervisor's reports on observed performance (in the workplace): practicing aseptic techniques as appropriate to the case and setting, investigating and managing infection, prescribing antibiotics according to guidelines
- Completion of infection control induction in the workplace
- Personal Protective Equipment Training Course (In hospital)

Self-Care and Maintaining Well-Being

Objectives:

1. To ensure that trainees understand how their personal histories and current personal lives, as well as their values, attitudes, and biases affect their care of patients so that they can use their emotional responses in patient care to their patients' benefit
2. To ensure that trainees care for themselves physically and emotionally, and seek opportunities for enhancing their self-awareness and personal growth

Medical Council Domains of Good Professional Practice: Patient Safety and Quality of Patient Care, Relating to Patients, Communication and Interpersonal Skills, Collaboration and Teamwork, Management (including self-management).

KNOWLEDGE

- Self-awareness including preferences and biases
- Personal psychological strengths and limitations
- Understand how personality characteristics, such as need for approval, judgemental tendencies, needs for perfection and control etc., affect relationships with patients and others
- Knowledge of core beliefs, ideals, and personal philosophies of life, and how these relate to own goals in medicine
- Know how family-of-origin, race, class, religion and gender issues have shaped own attitudes and abilities to discuss these issues with patients
- Understand the difference between feelings of sympathy and feelings of empathy
- Know the factors between a doctor and patient that enhance or interfere with abilities to experience and convey empathy
- Understanding of own attitudes toward uncertainty and risk taking and own need for reassurance
- How own relationships with certain patients can reflect attitudes toward paternalism, autonomy, benevolence, non-malfeasance and justice
- Recognise own feelings in straightforward and complex patient-doctor interactions
- Recognising the symptoms of stress and burn out

SKILLS

- Exhibiting empathy and showing consideration for all patients, their impairments and attitudes irrespective of cultural and other differences
- Ability to create boundaries with patients that allow for therapeutic alliance
- Challenge authority appropriately from a firm sense of own values and integrity and respond appropriately to situations that involve abuse, unethical behaviour and coercion
- Recognise own limits and seek appropriate support and consultation
- Work collaboratively and effectively with colleagues and other members of health care teams
- Manage effectively commitments to work and personal lives, taking the time to nurture important relationship and oneself
- Ability to recognise when falling behind and adjusting accordingly
- Demonstrating the ability to cope with changing circumstances, variable demand, being prepared to re-prioritise and ask for help
- Utilising a non-judgemental approach to patient's problem
- Recognise the warning signs of emotional ill-health in self and others and be able to ask for appropriate help
- Commitment to lifelong process of developing and fostering self-awareness, personal growth and well being
- Be open to receiving feedback from others as to how attitudes and behaviours are affecting their care of patients and their interactions with others
- Holding realistic expectations of own and of others' performance, time-conscious, punctual
- Valuing the breadth and depth of experience that can be accessed by associating with professional colleagues

ASSESSMENT & LEARNING METHODS

- On-going supervision
- RCPI Ethics programmes
- Wellness Matters Course
- RCPI HST Leadership in Clinical Practice course

Communication in Clinical and Professional Setting

Objective: To demonstrate the ability to communicate effectively and sensitively with patients, their relatives, carers and with professional colleagues in different situations.

Medical Council Domains of Good Professional Practice: Relating to Patients; Communication and Interpersonal Skills.

KNOWLEDGE

Within a consultation

- How to effectively listen and attend to patients
- How to structure an interview to obtain/convey information; identify concerns, expectations and priorities; promote understanding, reach conclusions; use appropriate language.
- How to empower the patient and encourage self-management

Difficult circumstances

- Understanding of potential areas for difficulty and awkward situations
- How to negotiate cultural, language barriers, dealing with sensory or psychological and/or intellectual impairments and how to deal with challenging or aggressive behaviour
- Knowing how and when to break bad news
- How to communicate essential information where difficulties exist, how to appropriately utilise the assistance of interpreters, chaperones, and relatives.
- How to deal with anger and frustration in self and others
- Selecting appropriate environment; seeking assistance, making and taking time

Dealing with professional colleagues and others

- How to communicate with doctors and other members of the healthcare team
- How to provide a concise, written, verbal, or electronic, problem-orientated statement of facts and opinions
- The legal context of status of records and reports, of data protection confidentiality
- Freedom of Information (FOI) issues
- Understanding of the importance of legible, accessible, records to continuity of care
- Knowing when urgent contact becomes necessary and the appropriate place for verbal, telephone, electronic, or written communication
- Recognition of roles and skills of other health professionals
- Awareness of own abilities/limitations and when to seek help or give assistance, advice to others; when to delegate responsibility and when to refer

Maintaining continuity of care

- Understanding the relevance of continuity of care to outcome, within and between phases of healthcare management
- The importance of completion of tasks and documentation, e.g. before handover to another team, department, specialty, including identifying outstanding issues and uncertainties
- Knowledge of the required attitudes, skills and behaviours which facilitate continuity of care including, being available and contactable, alerting others to avoid potential confusion or misunderstanding through communications failure

Giving explanations

- The importance of possessing the facts, and of recognising uncertainty and conflicting evidence on which decisions have to be based
- How to secure and retain attention avoiding distraction
- Understanding how adults receive information best, the relative value of the spoken, written, visual means of communication, use of reinforcement to assist retention
- Knowledge of the risks of information overload
- Tailoring the communication of information to the level of understanding of the recipient
- Strategies to achieve the level of understanding necessary to gain co-operation and partnership; compliance, informed choice, acceptance of opinion, advice, recommendation

Responding to complaints

- Value of hearing and dealing with complaints promptly; the appropriate level, the procedures (departmental and institutional); sources of advice, and assistance available
- The importance of obtaining and recording accurate and full information, seeking confirmation from multiple sources
- Knowledge of how to establish facts, identify issues and respond quickly and appropriately to a complaint received

SKILLS

- Ability to appropriately elicit facts, using a mix of open and closed-ended questions
- Using “active listening” techniques such as nodding and eye contact
- Giving information clearly, avoiding jargon, confirming understanding, ability to encourage co-operation, compliance; obtaining informed consent
- Showing consideration and respect for other’s culture, opinions, patient’s right to be informed and make choices
- Respecting another’s right to opinions and to accept or reject advice
- Valuing perspectives of others contributing to management decisions
- Conflict resolution
- Dealing with complaints
- Communicating decisions in a clear and thoughtful manner
- Presentation skills
- Maintaining (legible) records
- being available, contactable, time-conscious
- Setting realistic objectives, identifying and prioritising outstanding problems
- Using language, literature (e.g. leaflets) diagrams, educational aids and resources appropriately
- Establish facts, identify issues and respond quickly and appropriately to a complaint received
- Accepting responsibility, involving others, and consulting appropriately
- Obtaining informed consent
- Discussing informed consent
- Giving and receiving feedback

ASSESSMENT & LEARNING METHODS

- Mastering Communication course (Year 1)
- Consultant feedback at annual assessment
    - Workplace based assessment e.g. Mini-CEX, DOPS, CBD
    - Educational supervisor’s reports on observed performance (in the workplace): communication with others e.g. at handover. ward rounds, multidisciplinary team members
- Presentations
- RCPI Ethics programmes
- RCPI HST Leadership in Clinical Practice Course

Leadership

Objective: To have the knowledge, skills and attitudes to act in a leadership role and work with colleagues to plan, deliver and develop services for improved patient care and service delivery.

Medical Council Domains of Good Professional Practice: Patient Safety and Quality of Patient Care; Communication and Interpersonal Skill; Collaboration and Teamwork; Management (including Self-Management); Scholarship.

KNOWLEDGE

Personal qualities of leaders

- Knowledge of what leadership is in the context of the healthcare system appropriate to training level
- The importance of good communication in teams and the role of human interactions on effectiveness and patient safety

Working with others

- Awareness of own personal style and other styles and their impact on team performance
- The importance of good communication in teams and the role of human interactions on effectiveness and patient safety

Managing services

- The structure and function of Irish health care system
- Awareness of the challenges of managing in healthcare
    - Role of governance
    - Clinical directors
- Knowledge of planning and design of services
- Knowledge and understanding of the financing of the health service
    - Knowledge of how to prepare a budget
    - Defining value
    - Managing resources
- Knowledge and understanding of the importance of human factors in service delivery
    - How to manage staff training, development and education
- Managing performance
    - How to perform staff appraisal and deal effectively with poor staff performance
    - How to rewards and incentivise staff for quality and efficiency

Setting direction

- The external and internal drivers setting the context for change
- Knowledge of systems and resource management that guide service development
- How to make decisions using evidence-based medicine and performance measures
- How to evaluate the impact of change on health outcomes through ongoing service evaluation

SKILLS

- Effective communication with patients, families and colleagues
- Co-operation and collaboration with others; patients, service users, carers colleagues within and across systems
- Being an effective team player
- Ability to manage resources and people
- Managing performance and performance indicators

Demonstrating personal qualities

- Efficiently and effectively managing one-self and one's time especially when faced with challenging situations
- Continues personal and professional development through scholarship and further training and education where appropriate
- Acting with integrity and honesty with all people at all times
- Developing networks to expand knowledge and sphere of influence
- Building and maintaining key relationships
- Adapting style to work with different people and different situations
- Contributing to the planning and design of services

ASSESSMENT & LEARNING METHODS

- Mastering Communication course (Year 1)
- RCPI HST Leadership in Clinical Practice (Year 3 – 5)
- Consultant feedback at annual assessment
- Workplace based assessment e.g. Mini-CEX, DOPS, CBD
- Educational supervisor's reports on observed performance (in the workplace): on management and leadership skills
- Involvement in hospital committees where possible e.g. Division of Medicine, Drugs and Therapeutics, Infection Control etc.

Quality Improvement

Objective: To demonstrate the ability to identify areas for improvement and implement basic quality improvement skills and knowledge to improve patient safety and quality in the healthcare system.

Medical Council Domains of Good Professional Practice: Patient Safety and Quality of Patient Care; Communication and Interpersonal Skills; Collaboration and Teamwork; Management; Relating to Patients; Professionalism

KNOWLEDGE

Personal qualities of leaders

- The importance of prioritising the patient and patient safety in all clinical activities and interactions

Managing services

- Knowledge of systems design and the role of microsystems
- Understanding of human factors and culture on patient safety and quality

Improving services

- How to ensure patient safety by adopting and incorporating a patient safety culture
- How to critically evaluate where services can be improved by measuring performance, and acting to improve quality standards where possible
- How to encourage a culture of improvement and innovation

Setting direction

- How to create a 'burning platform' and motivate other healthcare professionals to work together within quality improvement
- Knowledge of the wider healthcare system direction and how that may impact local organisations

SKILLS

- Improvement approach to all problems or issues
- Engaging colleagues, patients and the wider system to identify issues and implement improvements
- Use of quality improvement methodologies, tools and techniques within every day practice
- Ensuring patient safety by adopting and incorporating a patient safety culture
- Critically evaluating where services can be improved by measuring performance, and acting to raise standards where possible
- Encouraging a culture of improvement and innovation

Demonstrating personal qualities

- Encouraging contributions and involvement from others including patients, carers, members of the multidisciplinary team and the wider community
- Considering process and system design, contributing to the planning and design of services

ASSESSMENT & LEARNING METHODS

- RCPI HST Leadership in Clinical Practice
- Consultant feedback at annual assessment
- Involvement in hospital committees where possible e.g. Division of Medicine, Drugs and Therapeutics, Infection Control etc.

Scholarship

Objective: To develop skills in personal/professional development, teaching, educational supervision and research

Medical Council Domains of Good Professional Practice: Scholarship

KNOWLEDGE

Teaching, educational supervision and assessment

- Principles of adult learning, teaching and learning methods available and strategies
- Educational principles directing assessment methods including, formative vs. summative methods
- The value of regular appraisal / assessment in informing training process
- How to set effective educational objectives and map benefits to learner
- Design and delivery of an effective teaching event, both small and large group
- Use of appropriate technology / materials

Research, methodology and critical evaluation

- Designing and resourcing a research project
- Research methodology, valid statistical analysis, writing and publishing papers
- Ethical considerations and obtaining ethical approval
- Reviewing literature, framing questions, designing a project capable of providing an answer
- How to write results and conclusions, writing and/or presenting a paper
- How to present data in a clear, honest and critical fashion

Audit

- Basis for developing evidence-based medicine, kinds of evidence, evaluation; methodologies of clinical trials
- Sources from which useful data for audit can be obtained, the methods of collection, handling data, the audit cycle
- Means of determining best practice, preparing protocols, guidelines, evaluating their performance
- The importance of re-audit

SKILLS

- Bed-side undergraduate and post graduate teaching
- Developing and delivering lectures
- Carrying out research in an ethical and professional manner
- Performing an audit
- Presentation and writing skills – remaining impartial and objective
- Adequate preparation, timekeeping
- Using technology / materials

ASSESSMENT & LEARNING METHODS

- An Introduction to Health Research (online)
- Performing audit course (online)
- Effective Teaching and Supervising Skills course (online) - recommended
- Educational Assessment Skills course - recommended
- Health Research Methods for Clinicians - recommended

Management

Objective: To understand the organisation, regulation and structures of the health services, nationally and locally, and to be competent in the use and management of information on health and health services, to develop personal effectiveness and the skills applicable to the management of staff and activities within a healthcare team.

Medical Council Domains of Good Professional Practice: Management.

KNOWLEDGE

Health service structure, management and organisation

- The administrative structure of the Irish Health Service, services provided in Ireland and their funding and how to engage with these for best results
- Department of Health, HSE and hospital management structures and systems
- The national regulatory bodies, health agencies and patient representative groups
- Understanding the need for business plans, annual hospital budgets, the relationship between the hospital and PCCC

The provision and use of information in order to regulate and improve service provision

- Methods of collecting, analysing and presenting information relevant to the health of a population and the apportionment of healthcare resources
- The common ways in which data is presented, knowing of the sources which can provide information relevant to national or to local services and publications available

Maintaining medical knowledge with a view to delivering effective clinical care

- Understanding the contribution that current, accurate knowledge can make to establishing clinical effectiveness, best practice and treatment protocols
- Knowledge of sources providing updates, literature reviews and digests

Delegation skills, empowerment and conflict management

- How to assess and develop personal effectiveness, improve negotiating, influencing and leadership skills
- How to manage time efficiently, deal with pressure and stress
- How to motivate others and operate within a multidisciplinary team

SKILLS

- Chairing, organising and participating in effective meetings
- Managing risks
- Managing time
- Delegating tasks effectively
- Managing conflicts
- Exploring, directing and pursuing a project, negotiating through the relevant departments at an appropriate level
- Ability to achieve results through an understanding of the organisation and its operation
- Ability to seek / locate information in order to define an issue needing attention e.g. to provide data relevant to a proposal for change, establishing a priority, obtaining resources
- Ability to make use of information, use IT, undertake searches and obtain aggregated data, to critically evaluate proposals for change e.g. innovative treatments, new technologies
- Ability to adjust to change, apply management, negotiating skills to manage change
- Appropriately using management techniques and seeking to improve these skills and personal effectiveness

ASSESSMENT & LEARNING METHODS

- Mastering Communication course
- Performing audit course (online)
- RCPI HST Leadership in Clinical Practice
- Annual audit
- Consultant feedback on management and leadership skills
- Involvement in hospital committees

Standards of Care

Objective: To be able to consistently and effectively assess and treat patients' problems

Medical Council Domains of Good Professional Practice: Patient Safety and Quality of Patient Care; Relating to Patients; Communication and Interpersonal Skills; Collaboration and Teamwork: Management (including Self-Management); Clinical Skills.

KNOWLEDGE

Diagnosing Patients

- How to carry out appropriate history taking
- How to appropriately examine a patient
- How to make a differential diagnosis

Investigation, indications, risks, cost-effectiveness

- The pathophysiological basis of the investigation
- Understand the clinical significance of references ranges, positive and negative predictive value and potential risks of inappropriate tests
- The procedures for commonly used investigations, common or/and serious risks
- Understanding of the sensitivity and specificity of results, artefacts, PPV and NPV
- Understanding significance, interpreting and explaining results of investigations
- Logical approach in choosing, sequencing and prioritising investigations

Treatment and management of disease

- Natural history of diseases
- Quality of life concepts
- How to accurately assess patient's needs, prescribe, arrange treatment, recognise and deal with reactions / side effects
- How to set realistic therapeutic goals, to utilise rehabilitation services, and use palliative care approach appropriately
- Recognising that illness (especially chronic and/or incapacity) has an impact on relationships and family, having financial as well as social effects e.g. driving

Disease prevention and health education

- Screening for disease: methods, advantages and limitations
- Health promotion and support agencies; means of providing sources of information for patients
- Risk factors, preventive measures, and change strategies applicable to smoking, alcohol, drug abuse, and lifestyle
- Disease notification; methods of collection and sources of data

Notes, records, correspondence

- Functions of medical records, their value as an accurate up-to-date commentary and source of data
- An understanding of the need and appropriate use of problem-orientated discharge notes, letters, more detailed case reports, concise out-patient reports and focused reviews
- Appreciating the importance of up-to-date, easily available, accurate information, and the need for communicating promptly e.g. with primary care

Prioritising, resourcing and decision taking

- How to prioritise demands, respond to patients' needs and sequence urgent tasks
- Establishing (clinical) priorities e.g. for investigations, intervention; how to set realistic goals; understanding the need to allocate sufficient time, knowing when to seek help
- Understanding the need to complete tasks, reach a conclusion, make a decision, and take action within allocated time
- Knowing how and when to conclude

Handover

- Know what are the essential requirements to run an effective handover meeting
    - Sufficient and accurate patients information
    - Adequate time
    - Clear roles and leadership
    - Adequate IT
- Know how to prioritise patient safety
    - Identify most clinically unstable patients
    - Use ISBAR (Identify, Situation, Background, Assessment, Recommendations)
    - Proper identification of tasks and follow-ups required
    - Contingency plans in place
- Know how to focus the team on actions
    - Tasks are prioritised
    - Plans for further care are put in place
    - Unstable patients are reviewed

Relevance of professional bodies

- Understanding the relevance to practice of standards of care set down by recognised professional bodies – the Medical Council, Medical Colleges and their Faculties, and the additional support available from professional organisations e.g. IMO, Medical Defence Organisations and from the various specialist and learned societies

SKILLS

- Taking and analysing a clinical history and performing a reliable and appropriate examination, arriving at a diagnosis and a differential diagnosis
- Liaising, discussing and negotiating effectively with those undertaking the investigation
- Selecting investigations carefully and appropriately, considering (patients') needs, risks, value and cost effectiveness
- Appropriately selecting treatment and management of disease
- Discussing, planning and delivering care appropriate to patient's needs and wishes
- Preventing disease using the appropriate channels and providing appropriate health education and promotion
- Collating evidence, summarising, recognising when objective has been met
- Screening
- Working effectively with others including
    - Effective listening
    - Ability to articulate and deliver instructions
    - Encourage questions and openness
    - Leadership skills
- Ability to prioritise
- Ability to delegate effectively
- Ability to advise on and promote lifestyle change, stopping smoking, control of alcohol intake, exercise and nutrition
- Ability to assess and explain risk, encourage positive behaviours e.g. immunisation and preventive measures
- Involve patients' in solving their health problems, by providing information and education
- Availing of support provided by voluntary agencies and patient support groups, as well as expert services e.g. detoxification / psychiatric services
- Act in accordance with, up to date standards on palliative care needs assessment
- Valuing contributions of health education and disease prevention to health in a community
- Compile accurate and appropriate detailed medical notes and care reports including the results of examinations, investigations, procedures performed, sufficient to provide an accurate, detailed account of the diagnostic and management process and outcome, providing concise, informative progress reports (both written and oral)
- Transfer information in an appropriate and timely manner

- Maintaining legible records in line with the Guide to Professional Conduct and Ethics for Registered Medical Practitioners in Ireland
- Actively engaging with professional/representative/specialist bodies

ASSESSMENT & LEARNING METHODS

- Consultant feedback
- Workplace based assessment e.g. Mini-CEX, DOPS, CBD
- Educational supervisor's reports on observed performance (in the workplace)
- Annual Audit
- Medical Council Guide to Professional Conduct and Ethics

Dealing with & Managing Acutely Ill Patients in Appropriate Specialties

Objectives: To be able to assess and initiate management of patients presenting as emergencies, and to appropriately communicate the diagnosis and prognosis. Trainees should be able to recognise the critically ill and immediately assess and resuscitate if necessary, formulate a differential diagnosis, treat and/or refer as appropriate, elect relevant investigations and accurately interpret reports.

Medical Council Domains of Good Professional Practice: Patient Safety and Quality of Patient Care, Clinical Skills.

KNOWLEDGE

Management of acutely ill patients with medical problems

- Presentation of potentially life-threatening problems
- Indications for urgent intervention, the additional information necessary to support action (e.g. results of investigations) and treatment protocols
- When to seek help, refer/transfer to another specialty
- ACLS protocols
- Ethical and legal principles relevant to resuscitation and DNAR in line with National Consent Policy
- How to manage acute medical intake, receive and refer patients appropriately, interact efficiently and effectively with other members of the medical team, accept/undertake responsibility appropriately
- Management of overdose
- How to anticipate / recognise, assess and manage life-threatening emergencies, recognise significantly abnormal physiology e.g. dysrhythmia and provide the means to correct e.g. defibrillation
- How to convey essential information quickly to relevant personnel: maintaining legible up-to-date records documenting results of investigations, making lists of problems dealt with or remaining, identifying areas of uncertainty; ensuring safe handover

Managing the deteriorating patient

- How to categorise a patients' severity of illness using Early Warning Scores (EWS) guidelines
- How to perform an early detection of patient deterioration
- How to use a structured communication tool (ISBAR)
- How to promote an early medical review, prompted by specific trigger points
- How to use a definitive escalation plan

Discharge planning

- Knowledge of patient pathways
- How to distinguish between illness and disease, disability and dependency
- Understanding the potential impact of illness and impairment on activities of daily living, family relationships, status, independence, awareness of quality of life issues
- Role and skills of other members of the healthcare team, how to devise and deliver a care package
- The support available from other agencies e.g. specialist nurses, social workers, community care
- Principles of shared care with the general practitioner service
- Awareness of the pressures/dynamics within a family, the economic factors delaying discharge but recognise the limit to benefit derived from in-patient care

SKILLS

- BLS/ACLS (or APLS for Paediatrics)
- Dealing with common medical emergencies
- Interpreting blood results, ECG/Rhythm strips, chest X-Ray, CT brain
- Giving clear instructions to both medical and hospital staff
- Ordering relevant follow up investigations
- Discharge planning, including complex discharge
- Knowledge of HIPE (Hospital In-Patient Enquiry)
- Multidisciplinary team working
- Communication skills
- Delivering early, regular and on-going consultation with family members (with the patient's permission) and primary care physicians
- Remaining calm, delegating appropriately, ensuring good communication
- Attempting to meet patients'/ relatives' needs and concerns, respecting their views and right to be informed in accordance with Medical Council Guidelines
- Establishing liaison with family and community care, primary care, communicate / report to agencies involved
- Demonstrating awareness of the wide ranging effects of illness and the need to bridge the gap between hospital and home
- Categorising a patients' severity of illness
- Performing an early detection of patient deterioration
- Use of structured communication tools (e.g. ISBAR)

ASSESSMENT & LEARNING METHODS

- ACLS course
- Record of on call experience
- Mini-CEX (acute setting)
- Case Based Discussion (CBD)
- Consultant feedback

Therapeutics and Safe Prescribing

Objective: To progressively develop ability to prescribe, review and monitor appropriate therapeutic interventions relevant to clinical practice in specific specialities including non-pharmacological therapies and preventative care.

Medical Council Domains of Good Professional Practice: Patient Safety and Quality of Patient Care.

KNOWLEDGE

- Pharmacology, therapeutics of treatments prescribed, choice of routes of administration, dosing schedules, compliance strategies; the objectives, risks and complications of treatment cost-effectiveness
- Indications, contraindications, side effects, drug interaction, dosage and route of administration of commonly used drugs
- Commonly prescribed medications
- Adverse drug reactions to commonly used drugs, including complementary medicines
- Identifying common prescribing hazards
- Identifying high risk medications
- Drugs requiring therapeutic drug monitoring and interpretation of results
- The effects of age, body size, organ dysfunction and concurrent illness or physiological state e.g. pregnancy on drug distribution and metabolism relevant to own practice
- Recognising the roles of regulatory agencies involved in drug use, monitoring and licensing e.g. IMB , and hospital formulary committees
- Procedure for monitoring, managing and reporting adverse drug reaction
- Effects of medications on patient activities including potential effects on a patient's fitness to drive
- The role of The National Medicines Information Centre (NMIC) in promoting safe and efficient use of medicine
- Differentiating drug allergy from drug side effects
- Know the difference between an early and late drug allergy, and drug side-effects
- Good Clinical Practice guidelines for seeing and managing patients who are on clinical research trials
- Best practice in the pharmacological management of cancer pain
- The management of constipation in adult patients receiving palliative care

SKILLS

- Writing a prescription in line with guidelines
- Appropriately prescribing for the elderly, children and pregnant and breast feeding women
- Making appropriate dose adjustments following therapeutic drug monitoring, or physiological change (e.g. deteriorating renal function)
- Reviewing and revising patients' long term medications
- Anticipating and avoiding defined drug interactions, including complementary medicines
- Advising patients (and carers) about important interactions and adverse drug effects including effects on driving
- Providing comprehensible explanations to the patient, and carers when relevant, for the use of medicines
- Being open to advice and input from other health professionals on prescribing
- Participating in adverse drug event reporting
- Take and record an accurate drug allergy history and history of previous side effects

ASSESSMENT & LEARNING METHODS

- Consultant feedback
- Workplace based assessment e.g. Mini-CEX, DOPS, CBD
- Educational supervisor's reports on observed performance (in the workplace): prioritisation of patient safety in prescribing practice
- Guidance for health and social care providers - Principles of good practice in medication reconciliation (HIQA)

Specialty Section

Clinical Genetics

Objectives: By the end of the educational programme, trainees must have the requisite knowledge, skills and attitudes listed in the curriculum, to diagnose and manage genetic aspects of a wide range of disorders in the following categories, including but not restricted to the conditions specified.

KNOWLEDGE

Cancers

- Common familial cancers – breast, ovary, bowel
- Rare genetic cancer syndromes – adenomatous polyposis coli, multiple endocrine neoplasia, NF 2, von Hippel Lindau disease.

Cardiac disorders

- Hereditary cardiomyopathies and conduction defects

Connective tissue disorders

- Marfan syndrome, Ehlers Danlos syndrome

Congenital abnormalities

- Single and multiple; malformations, deformations and disruptions; fetal and neonatal presentations
- Dysmorphic syndromes – common syndromes as well as some experience with rare disorders
- Learning disability – familial and syndromic causes

Chromosomal disorders –

- sporadic and familial; numerical and structural abnormalities

Single gene disorders

- Cystic fibrosis
- Deafness – isolated and syndromic deafness
- Fragile X syndrome – and other X-linked mental retardation syndromes
- Haematological disorders – haemoglobinopathies, haemophilia, thrombophilia
- Huntington disease – and other adult onset hereditary neurodegenerative disorders
- Inborn errors of metabolism
- Neurogenetic disorders – Spinal muscular atrophy, spinocerebellar ataxias, hereditary neuropathies, hereditary spastic paraplegia
- Neuromuscular disorders – myotonic dystrophy, Duchenne, Becker and others
- Neurocutaneous syndromes – neurofibromatosis 1 and tuberous sclerosis
- Ophthalmic genetic disorders – retinitis pigmentosa
- Renal disorders –adult and infantile polycystic kidney disease
- Skeletal dysplasias – achondroplasia, osteogenesis imperfecta, spondyloepiphyseal dysplasias
- Mitochondrial cytopathies – mitochondrial myopathies/encephalomyopathies and Leber's optic atrophy

Multifactorial disorders

- neural tube defects, epilepsies and common adult onset disorders
- Pharmacogenetic disorders – malignant hyperthermia and glucose 6 phosphate dehydrogenase deficiency
- Teratogens – alcohol and anticonvulsants

Ethical issues in Clinical genetics

- Predictive testing
- Testing of children

SKILLS

- Record and analyse family history data
- Obtain the medical history and carry out clinical examination as it relates to genetic diseases
- Diagnose genetic disease using clinical evaluation and genetic testing
- Choose appropriate investigations and interpret results
- Provide accurate information and effective genetic counselling to individuals and families
- Write clear summaries of genetic clinic consultations in post-clinic letters to colleagues and patients
- Formulate management plans for genetic aspects of genetic/hereditary disorders
- Perform risk calculation, including Bayes theorem
- Carry out phlebotomy, skin biopsy, and photography
- Conduct literature searches and use medical genetic databases
- Store and retrieve genetic data in single-disease genetic registers
- Work effectively in a team with other colleagues providing genetic services
- Liaise appropriately with colleagues from other specialists,
- Make use of lay organisations to support patients and families with genetic diseases
- Communicate and explain genetic issues to colleagues and the lay public
- Work effectively with colleagues in other disciplines

ASSESSMENT & LEARNING METHODS

- On-the-job training
- Personal study
- Dysmorphology Group Meetings
- Audit
- DOPS: Skin biopsy; buccal swab

History, Examination, Investigations, Management & Note Keeping Skills for Clinical Genetics

Objective: To be able to establish genetic diagnoses by means of clinical history taking, physical examination and use of appropriate investigations and to provide clinical genetic management for patients and families

KNOWLEDGE

History

- Knowledge on how to draw complex pedigrees accurately, including consanguinity loops, recording appropriate information

Investigations

- Surface anatomy
- Pitfalls in single gene inheritance including variable expressivity and reduced penetrance, somatic and gonadal mosaicism
- Differential diagnoses for genetic disorders
- Genetic databases and registers for information retrieval

Note keeping, letters etc

- Structure, function and legal implications of medical records & medico-legal reports.
- Know the relevance of data protection legislation pertaining to patient confidentiality

SKILLS

- Calculating genetic risk in single gene disorders by hand and by use of a computer programme
- Present genetic information to a patient in a sensitive and understanding manner
- Attention to detail and accuracy in collecting and checking family history and medical data
- Appreciate the confidentiality and ethical issues arising from family history gathering
- Clinical history taking, physical examination and use of appropriate investigations
- Provide clinical genetic management for patients and families

ASSESSMENT METHODS

- Audit (each year)
- Present difficult cases at weekly clinical meeting
- Record 10 pedigree cases
- Performing Audit course

Formal Genetics/Genomics and Basic Sciences

Objectives:

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

KNOWLEDGE

- Knowledge of:
- Knowledge of genomic technology along with its limitations
-
    - The chromosomal basis of heredity (mitosis and meiosis)
    - Mechanisms of origin of numerical and structural chromosome abnormalities
    - Behaviour of structural chromosome abnormalities at meiosis
    - The chemical structure of DNA and replication
    - Central dogma of cell biology: transcription and translation
    - History of genetics
- Modes of inheritance (Mendelian and non Mendelian)
- Risk calculations including combinatorial probability and Bayes Theorem
- The clinical embryology and molecular mechanisms of human malformation syndromes
- Principles of teratogenesis and pregnancy associated risks
- Mechanisms of mutagenesis and estimation of mutation rates
- Identification and critical evaluation of information
- Understanding of risks of over and under interpretation of variants

SKILLS

- Use primary sources of data
- Appreciation the impact of genetic disorders on individuals and families
- Appreciate potential benefits and harm of new genetic technologies
- Appreciate public concerns about the application of new genetic technologies e.g. NIPD/NIPT/CRISPR
- Recognition of different inheritance patterns in pedigrees
- Pedigree-based calculation of segregation ratios for structural chromosome abnormalities
- Empiric risk calculations (occurrence and recurrence risks)
- Perform Bayesian risk calculations including linkage-based risk calculations
- Calculate gene frequencies - the Hardy-Weinberg equilibrium and chi square tests of departure
- Apply knowledge to interpret results of chromosome and molecular genetic analysis
- Interpret variants of unknown significance (VUS)

ASSESSMENT & LEARNING METHODS

- Case based Discussion: Variation Interpretation Assessments
- Case presentations at Grand Rounds
- Study Day - relevant to key curricula items
- Attend mandated genomics courses
- Study Day - appropriate genetic courses e.g. mechanisms of origin of numerical and structural chromosome abnormalities
- Attend 12 x monthly multidisciplinary variant interpretation meetings

Common Genetic Referrals

Objectives: To provide the trainee with the skills and knowledge to be able to carry out specialist diagnosis, assessment and genetic counselling for the conditions previously listed.

KNOWLEDGE

- The genetic basis and clinical features of common genetic condition including Cystic Fibrosis, Down's syndrome, Fragile X, an x-linked recessive genetic condition
- The medical and surgical complications of common genetic conditions and indications for referral for specialist opinion
- Knowledge of long term complications of genetic conditions
- Molecular /cytogenetic testing and its application to diagnosis, predictive testing, carrier testing and prenatal diagnosis
- Application and limitations of current tests
- Knowledge of current clinical treatments for 'core' conditions and gene therapy trials

SKILLS

- Appreciate role of patient education, e.g. in type 1 neurofibromatosis
- Appreciate the role of the general practitioner in management of chronic disease
- Appreciate the role of support groups and be willing to provide appropriate information
- Apply good clinical care and counselling skills
- Be able to take a relevant history, perform an appropriate examination and formulate clinical diagnoses
- Be able to assess patients and families affected by genetic conditions
- Judge when it is necessary to sustain supportive relationships with patients with chronic disease
- Work in a team to develop and implement long term management utilising evidence based medicine and care pathways
- Be able to discuss reproductive options (AID, ICSI, IVF, pre-implantation diagnosis) with the patient and their partner in a sensitive manner
- Be able to discuss and formulate management plans with individuals/families
- Understand when predictive testing is appropriate to offer and steps you have to take to prepare a patient before undergoing a predictive test

ASSESSMENT & LEARNING METHODS

- Record up to five patient sessions by genetic counsellor in common genetic conditions per year in years 1 & 2
- 30 Cancer cases
- 30 Developmental delay cases
- 15 cases of common genetic conditions e.g. Cystic Fibrosis, sickle cell anaemia, chromosomal abnormality, an x-linked recessive genetic condition
- 10 adult neurology cases

Neurogenetics

Objectives: To provide the trainee with the skills and knowledge to recognise genetic causes of central and peripheral nervous system dysfunction

KNOWLEDGE

- Classification and molecular basis of common genetic neuromuscular disorders
- Predictive testing
- Genetic aspects and clinical presentation of trinucleotide repeat disorders
- Basic neuropathology and differential diagnosis of hereditary dementias
- Mitochondrial diseases – clinical, biochemical and genetic features
- Genetic causes of intellectual disability (static and progressive)
- Genetic contribution to autism and autistic spectrum disorders
- Genetic contribution to psychiatric disease in adults
- Huntington's

SKILLS

- Appreciation of family stresses caused by risk or eventuality of neurodegeneration
- Appreciate social problems encountered by adults with mild/moderate learning disability
- Understanding anticipation in relation to neurogenetic disease
- Recognise family history data that suggest familial neurological disease
- Verify diagnoses from old hospital records
- Be able to confirm clinical signs in affected individuals in the common disorders
- Be able to draw up a differential diagnosis and institute appropriate genetic testing
- Assessment of symptoms and signs in patients at risk of adult-onset neurogenetic disease
- Application of protocols for pre-symptomatic diagnosis of Huntington’s disease and other neurodegenerative disorders
- Make timely, appropriate referrals to other specialists such as neurologists, psychologists, psychiatrists, speech therapists
- Appreciate issues involved in predictive testing

ASSESSMENT & LEARNING METHODS

- Attend cross city paediatric neurology meetings
- Sit in on 3 Huntington's cases
- Sit in on 3 paediatric neurology clinics

Paediatric Genetics and Dysmorphology

Objectives: To provide the trainee with the skills and knowledge to make syndrome diagnosis in children

KNOWLEDGE

- Normal developmental milestones and diagnose delayed development
- Morphogenesis in terms of deformation, malformation, disruption and dysplasia
- Syndrome identification
- Common and rarer dysmorphic syndromes

SKILLS

- Recognise importance of clinical judgement, timing, and tact when diagnosing and informing parents of an infant with serious malformation or disability
- Appreciate the emotional reactions of parents following early diagnosis of syndrome or recognition of developmental delay
- Appreciate the adverse reaction families may experience following retraction of a previous diagnosis
- Have a rational approach to investigation of children with delayed development and/or dysmorphic syndromes
- Formulate differential diagnoses of unknown syndromes
- Cultivate critical assessment of database information and case reports to identify uncertainty and subjectivity in syndrome diagnosis
- Be able to provide a diagnostic service within a multidisciplinary clinical team
- Refer patients appropriately to specialist medical and surgical services
- Be able to use the Face2Gene (previously London dysmorphology) data base
- Variant interpretation in the context of rare disease
- Comment on 10 external cases over the programme.

ASSESSMENT & LEARNING METHODS

- Present known and unknown cases
- Present at Belfast or ISHG or at dysmorphology meeting twice during training
- Write up case report of case

Cardiac Disorders

Objectives:

- Demonstrate the ability to diagnose inherited cardiac conditions (ICC)
- Demonstrate the ability to recommend targeted screening in individuals who are identified as having increased risk of an ICC
- Demonstrate the ability to coordinate appropriate molecular genetic testing

KNOWLEDGE

- Classification and molecular basis of common ICC syndromes
- Knowledge of clinical features of ICC syndromes, including Marfan, Loeys-Dietz syndrome and related disorders
- Current recommendations concerning cardiac surveillance in ICC families
- Understand the impact of ICC risk on individuals and families
- Knowledge of genetic causes of sudden adult death
    1. Hypertrophic cardiomyopathy
    2. Long QT
    3. ARVC
    4. CPVT
    5. Brugada

SKILLS

- Be able to take a relevant history, perform an appropriate examination
- Work with bereaved families following sudden adult death
- Use of Ghent criteria for diagnosing Marfan syndrome
- Assessment of screening protocols for at-risk relatives
- Coordinate diagnostic genetic testing in ICC families
- Be aware of process involved in Predictive testing
- Identify at-risk patients and relatives who are eligible to participate in prevention strategies (e.g. therapeutic trials)
- Demonstrate awareness of the roles of primary care, specialist nurses and genetic counsellors and their importance in assessing families where relatives are at risk of developing ICC
- Inform patients about lifestyle factors that affect risk
- Support primary and secondary care professionals with the long-term management of selected patients with ICC syndromes
- Demonstrate awareness of psychological impact of sudden adult death
- Interpret Variants of Unknown Significance (VOUS)

ASSESSMENT & LEARNING METHODS

- Present known and unknown cases
- Present at Belfast or ISHG meeting twice during training
- Write up case report of case
- Attendance at multidisciplinary cardiac genetics meetings

Cancer Genetics

Objectives:

- Trainee is able to diagnose rare paediatric and adult cancer syndromes and recognise when common cancers are likely to have a single gene basis
- The trainee can recommend targeted screening in individuals who are identified as having increased risk
- Trainee can coordinate appropriate molecular genetic testing

KNOWLEDGE

- The genetic and environmental factors that affect risk of developing cancer
- Current recommendations concerning tumour surveillance in cancer
- Knowledge of clinical features of genetic cancer syndromes
- Knowledge of DNA repair disorders
- Genetic mechanisms in neoplasia: Knudson's two-hit hypothesis, oncogenes
- Knowledge of molecular basis of cancer genetic syndromes
- Knowledge of cancer registers and other sources to verify diagnoses
- Knowledge of disease registers (e.g. von Hippel Lindau disease) to support follow-up of affected and at-risk patients
- Screening protocols for at-risk relatives
- Mechanistic tools for calculating likelihood of cancer being inherited
- Understanding how genetic diagnoses can influence treatment and management

SKILLS

- Demonstrate awareness of the roles primary care and genetic associates play in assessing families where relatives are at risk of developing cancer
- Inform patients about lifestyle factors that affect cancer risk
- Support general practitioners with the long-term management of selected patients with familial cancer syndromes
- Liaise with other specialists as appropriate e.g. for advice about prophylactic mastectomy and work as a member of a multidisciplinary team
- Identify high risk family from a questionnaire
- Testing risk prediction algorithms
- Understand the impact of cancer risk on individuals and families
- Identify at-risk patients and relatives who are eligible to participate in trials of cancer prevention strategies
- Be able to identify high risk family from a questionnaire
- Interpret Variant of Unknown Significance (US)

ASSESSMENT & LEARNING METHODS

- Requirements for training: 6 months working exclusively on families referred for cancer risk assessment
- Study day: Cancer module (with oncology SpRs or appropriate online course or cross city meeting with cancer genetics (St James'))

Prenatal Diagnosis and Neonatal Dysmorphology

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

KNOWLEDGE

- Process and limitations of clinical and laboratory diagnostic procedures at neonatal post mortem examination
- Knowledge of guidelines on retention and storage of fetal tissues
- Know the natural history of prenatally diagnosed conditions including autosomal and sex chromosome aneuploidy syndromes
- Knowledge of the Irish legal framework pertaining to termination of pregnancy
- Knowledge of Council of Europe Guidelines on Tissue storage
- Knowledge of Non-invasive Pre-natal Testing (NIPT) and Non-Invasive Pre-natal Diagnosis (NIPD)
- Knowledge of the role of NIPD/NIPT in terms of antenatal testing
- Knowledge of the differences between NIPD and NIPT
- Knowledge of PIGD

SKILLS

- Appreciate the different perspectives on advantages and disadvantages of prenatal diagnosis in each situation
- Non-judgmental appreciation of the ethical and religious dimensions to prenatal diagnosis
- Awareness of the adverse psychological effects of termination of pregnancy for fetal abnormality
- Interpret family history data and trace old medical records
- Perform post-mortem clinical analysis of the neonate (examination, measurements, photography, radiology, tissue sampling and storage for diagnostic studies)
- Use syndrome databases in syndrome diagnosis
- Provide genetic advice for women who may undergo prenatal diagnosis
- Assess clinical significance of chromosome, DNA, and fetal imaging studies in the context of fetal abnormality or risk thereof
- Formulate differential diagnoses and assess prognosis in collaboration with the fetal medicine team
- Perform risk-assessment when pregnancies are exposed to hazards such as congenital infections, alcohol, ionising irradiation or drugs
- Sensitive disclosure of abnormal test results or diagnoses in the antenatal period
- Ability to organise an NIPD for an at risk couple
- Understand the sensitivity & specificity of test

ASSESSMENT & LEARNING METHODS

- Attendances at neonatal post-mortem examination,
- Study day – attendance at quarterly joint fetal medicine meetings
- attend a minimum of 3 half day sessions in fetal medicine unit to observe the following procedures: amniocentesis and chorionic villus sampling and ultrasound scanning of pregnancies.
- Attendance at multidisciplinary fetal medicine meetings

Biochemical Genetics & Metabolic Diseases

Objectives:

- To become competent with the diagnosis, treatment and follow up of patients with common Hereditary Metabolic Diseases (HMDs).
- To become familiar with the management of patients in acute metabolic crisis and also with the multidisciplinary care required for patient with chronic diseases, including psychosocial care.
- To understand the application of Genomic and Precision Medicine to Inherited Metabolic Disorders

KNOWLEDGE

- Understand basic physiology & biochemistry including fluid and electrolyte balance
- Understand metabolic response to fasting, lactate, ammonia, amino, organic & fatty acids
- Understand oxidative phosphorylation, lysosomal and peroxisomal metabolism
- Galactose & pathophysiology in Galactosemia
- Glucose lactate profile and lactate/pyruvate ratios
- Understand cholesterol and steroid metabolism
- Metabolic functional studies: including lactate/pyruvate profiling, fasting studies, & investigation of hyperammonaemia
- Biochemical Genetic/Genomic tests required in the investigation of developmental delay/intellectual disability
- Drug management & experience of drugs used in the treatment of metabolic intoxication
- The principles of dialysis for metabolic intoxication
- The applications of liver, HSCT and stem cell transplantation
- Gene/Genotype specific treatments in the area *e.g.* chaperone therapies in Fabry disease
- The principles of gene therapy and genome editing technologies
- The general nutritional parameters & the use of nutritional unwell & unstable diet regimes
- The parameters used to measure normal intellectual and psychological development, the assessment of IQ, behaviour and neuro psychological function
- The applications of clinical research
- Guidelines for investigation & management of Fabry Disease, MPSI, MPSII, MPSVI and be familiar with the Registries and outcome analyses

SKILLS

- To be familiar with Enzyme Replacement Therapy protocols for Lysosomal Storage Diseases & other therapies e.g. chaperones, substrate inhibition
- To be familiar with how Next Generation Sequencing is applied within the area of Biochemical Genetics
- To be familiar with the principles of gene therapy
- To be familiar with the interpretation of specialist biochemical testing, including plasma amino & urine organic acid analysis, acylcarnitine profiles, mitochondrial respiratory chain enzymology & lysosomal screening enzymology tests.
- To become familiar with the post mortem metabolic genetic autopsy.
- To become familiar with the principles of Newborn Screening, the Irish and European practice and understand the different opportunities of genetic screening.
- To perform one audit/review during the rotation

ASSESSMENT AND LEARNING METHODS

- Clinic attendance for 6 months (including paediatric (if possible) & adult & maternal PKU)
- The trainee will become familiar with clinical research trials and GCP
- 5 days attendance as observer at Newborn Screening Laboratory & one week immersion in laboratory to observe performance & analysis of amino & organic acid analysis Observe analysis of urinary GAGs
- Attend
    - Grand Rounds
    - Laboratory meeting
    - Metabolic Journal Club (with presentation at least one during the rotation)
    - Psycho-social meetings and selected pre-clinic meetings
- 40 cases during rotation to include 20 patients with Lysosomal Storage Diseases
- Provide teaching session in genetics to metabolic trainees and staff

Laboratory Genetics

Objective: The trainee acquires skills and knowledge to interpret genetic laboratory results within a clinical setting, by completing an attachment in the genetic laboratories

KNOWLEDGE

- Techniques for conventional chromosome analysis in different tissues
- Laboratory techniques and application of new cytogenetic tests e.g. Array CGH, FISH
- Use of ISCN nomenclature
- Molecular genetic techniques in common usage– (DNA extraction, Southern Blotting, PCR, DNA sequencing)
- Exome and genome sequencing : an understanding of the principles and practice of exome and genome sequencing
- Next generation sequencing (exomic & genomic) sequencing
- Application of DNA-based testing for gene mapping, linkage and mutation detection.
- Potential application of new DNA technologies
- Sensitivity and specificity of laboratory tests
- Use of DNA and molecular cytogenetic methods in pre implantation diagnosis
- The operation of national CF newborn screening programme
- Pre implantation genetics diagnosis
- Interpretation of clinical consequences of abnormal karyotypes and molecular test results
- Incidentals, awareness of how and where these arise and how to manage them
- Awareness of ESHG document on incidental findings
- Non-invasive prenatal testing and non-invasive prenatal screening; cell-free fetal DNA (cffDNA): an understanding of the sensitivity, specificity and positive predictive value of cffDNA for aneuploidy screening
- Understand techniques for conventional cytogenetic analysis in different tissues
- Interpret clinical consequences of chromosome rearrangements
- Understand the principles of FISH analysis and its applications
- Apply array-CGH in different clinical settings and interpret of CNV’s (including use of databases such as DECIPHER)
- Use ISCN nomenclature correctly
- Know the molecular genetic techniques in common usage: DNA extraction, Southern blotting, PCR, MLPA, and Sanger sequencing
- Understand the principles and application of next generation sequencing (NGS) technologies including targeted panels, clinical exome sequencing, whole exome sequencing, whole genome sequencing
- Interpret the large data set created from NGS using basic bioinformatics, filtering techniques, clinical and functional data
- Know OMIC technologies and their current and future applications
- Be aware of the Human Genome Variation (HGVS) nomenclature for single gene variants
- Understand the sensitivity and specificity of laboratory tests
- Investigate inborn errors of metabolism through liaison with metabolic disease colleagues and the genetic laboratory
- Be aware of the operation of local and national antenatal and newborn genetic disease screening programmes
- Knowledge and understanding of the ACCE wheel (CDC) and its implication for diagnostic testing
- To obtain a basic overview of proteomics/metabolomics and become aware of its clinical applications and how it is being incorporated into the field of medical/biochemical genetics

SKILLS

- Awareness of the importance of informed consent that arise in relation to storage of DNA samples and cell lines and ability to consent patients for storage and testing

- Willingness to liaise with colleagues to interpret laboratory results
- Liaise with molecular and cytogenetics scientists in analysis of test results
- Provide advice to laboratory on the wording of reports to referring clinicians
- Genetic risk calculation based on laboratory test results
- Be aware of importance of Bioinformatics & be able to do database searches (Decipher & Ensembl)
- Awareness of possibility of incidental findings and the methods required to interpret variants of unknown significance
- Awareness of the need for confirmation of diagnosis where screening techniques are used
- Undertake genetic risk calculation based on laboratory test results (incorporation of genetic test results into Bayesian calculations)
- Interpret results of cytogenetic, molecular genetic and biochemical tests
- Use databases including ENSEMBL, USCS and locus-specific databases for interpretation of results
- Liaise with laboratory scientists and bioinformaticians in the analysis of test results
- Provide advice to genetic laboratory colleagues on the wording of reports to referring clinicians
- Develop awareness of the importance of informed consent in relation to storage of DNA and cell lines
- Be able to take informed consent when undertaking genomic analyses
- Demonstrate awareness of the potential for incidental findings in genomic analyses and the complexity of these from the patient perspective
- Recognise the importance and impact of genetic test results for families and communicate implications of results clearly to them
- Show willingness to liaise with colleagues to interpret laboratory results
- Be able to adapt to new techniques and tests as they arise and incorporate them into clinical practice appropriately
- Familiarity with the use of proteomics/metabolomics to identify the underlying pathology of a disease/syndrome.
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ASSESSMENT & LEARNING METHODS

- Two weeks in chromosome and DNA laboratory
- Case Based discussion: Unusual Cytogenetic and Molecular Genetic cases
- Attend a bioinformatics course

Organisation and Provision of Genetics Services for Populations

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

KNOWLEDGE

- The genetic characteristics in different populations, mutant gene frequencies and disease prevalence
- The factors that influence decisions to instigate programmes of population screening for genetic diseases
- Sensitivity, specificity, and predictive values of screening tests
- Knowledge of current screening programmes
- Knowledge of appropriate population-based registers

SKILLS

- Appreciate ethical and social dimensions to population screening
- Understand the central role of patient education
- Appreciate the value of specialised clinics (breast clinics, lipid and cardiovascular risk factor clinics)
- Encourage patients to adopt a healthier lifestyle with specific emphasis on risk factor avoidance and promotion of behaviours that reduce risk of developing disease
- Team-working with database managers, genetic associates and nurse specialists in:
    - ‘cascade screening’ and provision of genetic services for extended families with common single gene disorders (cystic fibrosis, Xp21 muscular dystrophy, fragile X syndrome, Huntington’s disease)
    - family based screening for individuals at high risk of developing cancer
    - contribute to the maintenance of departmental genetic register systems
- Be able to explain the benefits and consequences of screening programmes
- Be aware of neonatal screening programmes in EU

ASSESSMENT & LEARNING METHODS

- Study Day
- Ethics Programme

Clinical Liaisons

Objectives: To equip the trainee with skills and knowledge to provide genetic advice within multidisciplinary clinic settings

KNOWLEDGE

- Genetic contribution with other specialists including:
    - Child development (depending on prior experience)
    - Vision
    - Hearing
    - Endocrine
    - Skeletal dysplasia
    - Neurological
    - Cranio-facial malformation
    - Tumour surveillance
    - cardiac

SKILLS

- Team working skills
- Develop a special interest clinic
- Develop skills and liaisons needed to nurture new services, even in settings such as health centres or child development centres, outside of the genetics department

ASSESSMENT & LEARNING METHODS

- Attend multidisciplinary team meetings
- Case Based Discussion
- Attend three specialist NF and Cardiac Genetic specialty clinics

Patient Education and Disease Prevention

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

KNOWLEDGE

- Educating patients about:
  - disease
  - investigations
  - management
- Know disease course and manifestations
- Know investigation procedures including possible alternatives / choices
- Management strategies for genetic disease

Rare diseases

- Awareness of the EU recommendation on rare disease and how it impacts on patients with rare diseases from the Republic of Ireland.
- Awareness articles 12 & 13 of the EU cross border directive for travel for patients with rare diseases and the application of European Reference Networks
- Knowledge of the role of the Orphanet website and its use as a resource for patients with rare diseases and professionals who care for such patients
- Awareness of role of information office in uploading data on centres of expertise within Ireland
- Awareness of what is required from each expert centre before their data is allowed to be uploaded
- Awareness of the role of the National Rare disease office (NRDO) in Ireland and awareness of its use in highlighting new clinical trials

Environmental & lifestyle risk factors

- Understand the risk factors that may influence certain genetic diseases, including;
  - Life style
  - Smoking
  - Alcohol
  - Medication
- Knowledge of teratogenic potential of medication

Epidemiology & screening

- Know the methods of data collection and their limitations
- Know principles of 1o & 2o prevention & screening

SKILLS

- Assess an individual patient's risk factors.
- Encourage participation in appropriate disease prevention or screening programmes.
- Consider the:
  - positive & negative aspects of prevention
  - importance of patient confidentiality
- Give information to patients clearly in a manner that they can understand including written information
- Respect patient choice
- Consider involving patients in developing mutually acceptable investigation plans.
- Encourage patients to access:
  - further information
  - patient support groups
- Use Orphanet effectively to source information

ASSESSMENT & LEARNING METHODS

- Study Day
- Case Based Discussion
- Visit the NRDO

Use of Databases

Objective: To ensure that the trainee becomes proficient in the use of *Decipher* and *Ensembl* web based browser platforms (databases) for comparative genomics (human genome variants.)

KNOWLEDGE

- Be able to upload a case on to *Decipher*
- Be able to navigate through *Ensembl* and identify genes of interest within patients Copy Number Variation (CNV)
- Be able to investigate a variant and determine its pathogenicity or otherwise using *ClinVar* and *GnomAD* databases.
- Knowledge of international standardised processes used to determine pathogenicity of a variant.

SKILLS

- Identifying genes of interest and CNV's on databases
- Determination of the pathogenicity of a variant using appropriate methodologies and platforms
- Presentation of cases at variant meetings *e.g*. monthly variant meeting in OLCHC

ASSESSMENT & LEARNING METHODS

- DOPS x 3 on Use of Genetic Databases
- Record up to 50 cases per programme (Variant/CVV)
- Assessment of 8 cases by CBD

Documentation of Minimum Requirements for Training

- These are the minimum number of cases you are asked to document as part of your training. It is recommended you seek opportunities to attain a higher level of exposure as part of your self-directed learning and development of expertise.
- You should expect the demands of your post to exceed the minimum required number of cases documented for training.
- If you are having difficulty meeting a particular requirement, please contact your specialty coordinator

Curriculum Requirement	Required/Desirable	Minimum Requirement	Reporting Period	Form Name
Section 1 - Training Plan				
Personal Goals Plan (Copy of agreed Training Plan for your current training year signed by both Trainee & Trainer)	Required	1	Training Post	Clinical Activities
Section 2 - Training Activities				
Outpatient Clinics				Clinics
Biochemical genetics	Required	40	Training Programme	
Cystic Fibrosis	Required	3	Training Programme	
General Genetics	Required	88	Training Programme	
Neurofibromatosis/Tuberosclerosis/Neurogenetics	Required	6 (2 of each)	Training Programme	
Cardiac Genetics	Required	20	Training Programme	
Cancer Clinic (including breast/ovarian cancer clinics and Colorectal cancer clinics)	Required	60	Training Programme	
Adult Neurogenetics	Required	20	Training Programme	
Ophthalmology Genetics	Required	10	Training Programme	
Ward Rounds/Consultations				Clinical Activities
Consultations	Required	25	Year of Training	
Procedures/Practical Skills/Surgical Skills				Procedures, Skills, & DOPS
Skin Biopsy	Required	5	Training Programme	
Buccal Swabs	Required	5	Training Programme	
Additional/Special Experience Gained				Clinical Activities
Newborn screening laboratory	Required	1 (1 week)	Training Programme	
Chromosome and DNA laboratory (Cytogenetics and Molecular Genetics)	Required	2 x 2 weeks each	Training Programme	

Curriculum Requirement	Required/Desirable	Minimum Requirement	Reporting Period	Form Name
Time in Next Generation Sequencing Lab	Required	1 week	Training Programme	
Rare diseases network	Required	1 x 1 week		
Patient sessions with Genetic counselor (in 1st ***and*** 2nd Year; 5 per year)	Required	10	Training Programme	
Cancer Genetics (6 months' experience)	Required	1	Training Programme	
Cardiac Genetics (3 months' experience, including MDTs and 2 Cardiac clinics)	Required	1	Training Programme	
Time in fetal medicine unit (sessions)	Required	8	Training Programme	
Huntington's cases	Required	3	Training Programme	
Neonatal post-mortem examination (observe)	Desirable	1	Training Programme	
Record of cases				Cases
Cardiac	Required	5	Training Programme	
Ophthalmology	Required	5	Training Programme	
X-linked Pedigree cases	Required	10	Training Programme	
Rare Genetic Disease	Required	30	Training Programme	
Cancer cases (Including breast/ovarian cases & Colorectal cases)	Required	30	Training Programme	
Development delay / Learning Disability cases	Required	30	Training Programme	
Cases of common genetic conditions e.g. Cystic Fibrosis, sickle cell anaemia, chromosomal disorder an X-linked recessive genetic condition	Required	15	Training Programme	
Adult Neurology cases (Dementia/Ataxia/SCA/MND)	Required	10	Training Programme	
Lysosomal storage diseases (8 patients, 2 with known or suspected mitochondrial disease)	Required	8	Training Programme	
Pre-natal Diagnosis	Required	5	Training programme	
Paediatric Epilepsy	Required	5	Training Programme	
Birth Defects/Dysmorphology	Required	10	Training programme	
Inherited Hearing Loss	Required	5	Training Programme	
Traveller Population (Case Work up)	Required	3	Training Programme	
Management Experience	Desirable	1	Training Programme	Management Experience
Section 3 - Educational Activities				

Curriculum Requirement	Required/Desirable	Minimum Requirement	Reporting Period	Form Name
Courses				Teaching Attendance
Ethics Foundation	Required	1	Training Programme	
Ethics for General Medicine	Required	1	Training Programme	
An Introduction to Health Research	Required	1	Training Programme	
HST Leadership in Clinical Practice (before Year 3)	Required	1	Training Programme	
Mastering Communications (Year 1)	Required	1	Training Programme	
Performing Audit (Year 1)	Required	1	Training Programme	
ACLS	Required	1	Training Programme	
ESHG Recognised Online clinical bioinformatics course (to be agreed with Trainer)	Required	1	Training Programme	
St Georges Online Genomics	Optional	1	Training Programme	
Manchester Online Bioinformatics Course	Optional	1	Training Programme	
Bioinformatics course - Wellcome Trust Sanger Centre Cambridge	Desirable	1	Training Programme	
ACMG curated educational material for trainees	Desirable	1	Training Programme	
Educational material about NGS and genome sequencing and examples on the US Clingen site	Desirable	1	Training Programme	
Bertinoro ESHG course on genomics and NGS	Desirable	1	Training Programme	
RCPI “Cancer genetics”	Optional	1	Training Programme	
Wellness Matters	Required	1	Training Programme	
The fundamentals of GDPR (HSE land) (year 1)	Required	1	Training Programme	
Wessex Regional Genetics Laboratory Monthly variant Assessment Meeting	Desirable	12	Training Programme	
Study Days	Required	3	Year of Training	Teaching Attendance
Participation at In-house activities minimum of 1 per month from the categories below:				Attendance at Hospital Based Learning
Paediatric neurology meetings	Required	2	Year of Training	
Grand Rounds (present 2 in training programme)	Required	2	Training Programme	
Specialty Meeting – Neurology, endocrinology, dermatology	Required	6	Year of Training	
Dysmorphology group meetings	Required	2	Year of Training	
Other (minimum of 1 per month from the categories below:)				

Curriculum Requirement	Required/Desirable	Minimum Requirement	Reporting Period	Form Name
Journal Club	Required	2	Year of Training	
Pathology Conferences	Desirable	1	Year of Training	
MDT Meetings – attend at least two of each of the following and 30 per programme variant interpretation • Medicine (Cardiology, Neurology, Ophthalmology) • Tumor Board (Paediatrics) Crumlin • Cross City (Paediatrics) Neurology meeting • Paediatric Disorders of Sexual Development (Endocrine) • The Dublin Fetal Medicine group (3 per year) • Variant interpretation meetings	Required	50	Training Programme	
Examinations (European (UEMS) or British (KBA)) Recommended Year 3	Required	1	Training Programme	Examinations
Delivery of Teaching minimum 1 formal teaching session per year from the categories below: Lecture Tutorial	Desirable	1	Year of Training	Delivery of Teaching
Research Activities	Desirable	1	Training Programme	Research Activities
Audit Activities and Reporting (1 per year either to start or complete, Quality Improvement (QI) projects can be uploaded against audit)	Required	1	Year of Training	Audit & QI
Publications	Desirable	1	Year of Training	Additional Professional Experience
Presentations				Additional Professional Experience
A minimum of two presentations at genetics meetings and one general presentation	Required	3	Year of Training	
National/International meetings	Required	1	Year of Training	Additional Professional Experience
Committee Attendance	Desirable	1	Training Programme	Additional Professional Experience
Additional Qualifications	Desirable	1	Training Programme	Additional Professional Experience
Section 4 - Assessments				

Curriculum Requirement	Required/Desirable	Minimum Requirement	Reporting Period	Form Name
CBD	Required	5	Year of Training	Case Based Discussion
CBD - Variant Interpretation Assessment	Required	8	Year of Training	Case Based Discussion
DOPS				Procedures, Skills, & DOPS
Skin Biopsy	Required	3	Training Programme	
Buccal swab	Required	3	Training Programme	
Pedigree drawing (1st year)	Required	5	Training Programme	
Use of Genetic Databases	Required	3	Training Programme	
Mini-CEX (At least two Mini-CEX assessments)	Required	2	Year of Training	Mini-CEX
Quarterly Assessment/End of Post Assessment	Required	4	Year of Training	Quarterly/End of Post Assessment
End of Year Assessment	Required	1	Year of Training	End of Year Evaluation