INTERNATIONAL CLINICAL FELLOWSHIP TRAINING IN

CLINICAL GENETICS
This curriculum of training in Clinical Genetics was developed in 2014 and undergoes an annual review by Dr Sally Ann Lynch National Specialty Directors, Dr. Ann O'Shaughnessy, Head of Education and Professional Development and by the Clinical Genetics Training Committee. The curriculum is approved by the Irish Committee on Higher Medical Training.

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Introduction
The International Clinical Fellowship Programme (ICFP) provides a route for overseas doctors wishing to undergo structured and advanced postgraduate medical training in Ireland. The ICFP enables suitably qualified overseas postgraduate medical trainees to undertake a fixed period of active training in clinical services in Ireland. The programme is normally offered over one or two years of clinical training, after which the overseas doctors will be required to return to their country of origin. In limited certain circumstances, the period of training may extend to three years. The purpose of the ICFP is to enable overseas trainees to gain access to structured training and in active clinical environments that they cannot get in their own country, with a view to enhancing and improving the individual's medical training and learning and, in the medium to long term, the health services in their own countries. This Programme will allow participants to access a structured period of training and experience as developed by the Royal College of Physicians of Ireland to specifically meet the clinical needs of participants as defined by their home country's health service.

Aims
Upon satisfactory completion of the ICFP, the doctor will be competent to undertake comprehensive medical practice in their chosen specialty in a professional manner, in keeping with the needs of the healthcare system.

Competencies, at a level consistent with practice in the specialty, 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.
- Capability to be a scholar, contributing to development and research in the field of the chosen specialty.
- Professionalism.
- Ability to understand health care and identify and carry out system-based improvement of care.

Professionalism
Medical professionalism is a core element of being a good doctor. Good medical practice is based on a relationship of trust between profession and society, in which doctors are expected to meet the highest standards of professional practice and behaviour. It involves partnership between patient and doctor that is based on mutual respect, confidentiality, honesty, responsibility and accountability. In addition to maintaining clinical competence, a doctor should also:

- Show integrity, compassion and concern for others in day-to-day practice
- Develop and maintain a sensitive and understanding attitude with patients
- Exercise good judgement and communicate sound clinical advice to patients
- Search for the best evidence to guide professional practice
- Be committed to continuous improvement and excellence in the provision of health care whether working alone or as part of a team

Prior to commencing their sponsored clinical placements, all participants will also be required to undergo the mandatory screening requirements of the relevant clinical site/service including occupational health assessment and Garda/Police clearance.
Training Programme Duration & Organisation of Training

The period of clinical training that will be provided under the International Clinical Fellowship Programme (ICFP) is normally 12-24 months, after which the overseas doctors will be required to return to their country of origin. In certain circumstances, the period of training may extend to three years.

- Each ICFP is developed by the Royal College of Physicians of Ireland and will be specifically designed so as to meet the training needs of participants to support the health service in their home country.
- Appointees to the ICFP will be assessed by the Royal College of Physicians of Ireland to ensure that they possess the necessary requirements from a training and clinical service perspective.
- Each overseas doctor participating in the ICFP will be enrolled with the Royal College of Physicians of Ireland and will be under the supervision of a consultant doctor who is registered on the Specialist Division of the Register of Medical Practitioners maintained by the Medical Council and who is an approved consultant trainer.
- Appointees to the ICFP will normally be registered on the Supervised Division of the Register of Medical Practitioners maintained by the Medical Council in Ireland.
- Appointees will agree a training plan with their trainers at the beginning of each training year.
- For the duration of their International Medical Graduate (IMG) programme and associated clinical placements, all participants will remain directly employed and directly paid by their sponsoring state at a rate appropriate to their training level in Ireland and benchmarked against the salary scales applicable to NCHD’s in Ireland;
- Successful completion of an ICFP will result in the participant being issued with a formal Certificate of completion for the Fellowship Programme by the Royal College of Physicians of Ireland. This Certificate will enable the participant’s parent training body in their sponsoring home country to formally recognise and accredit their time spent training in Ireland.

The training programme offered will provide opportunities to fulfil all the requirements of the curriculum of training. There will be posts in both general hospitals and teaching hospitals. Each post within the programme will have a named trainer/educational supervisor and programmes will be under the direction of the National Specialist Director for the relevant medical specialty to be confirmed by the College. Programmes will be as flexible as possible consistent with curricular requirements, for example to allow the trainee to develop their sub-specialty interest.

ePortfolio logbook

Each trainee is responsible for maintaining an up-to-date record of progress through training and compiling a portfolio of achievements for presentation at each annual assessment review. 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. Up-to-date training records and an ePortfolio of achievements will be maintained by the trainee throughout. The training records 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 training plan. They will remain the property of the trainee and must be produced at their annual assessment review.

Trainees must co-operate with the College in completing their training plan. It is in a trainee’s own interest to maintain contact with the Royal College of Physicians of Ireland, and to respond promptly to all correspondence relating to training. At review, your ePortfolio will be examined.
Review

A consultant trainer/educational supervisor will be identified for each participant in the programme. He/she will be responsible for ensuring that the educational potential of the post is translated into effective training which is being fully utilized. Only departments approved for Training by the Royal College of Physicians of Ireland and its constituent training bodies will be used. The training objectives to be secured should be agreed between each 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. In each year trainees undergo a formal review by an appropriate panel. The panel will review in detail the training record, will explore with the trainee the range of experience and depth of understanding which has been achieved and consider individual trainer’s reports. An opportunity is also given to the trainee to comment on the training being provided; identifying in confidence any deficiencies in relation to a particular post. A quarterly and annual review of progress through training will be undertaken on behalf of the International Clinical Fellowship Programme (ICFP). These will include assessments and reports by educational supervisors, confirmation of achievements and the contents of the ePortfolio will be reviewed. At some or all of these annual reviews a non-specialty assessor will be present capable of addressing core competencies. The award of a Certificate of completion will be determined by a satisfactory outcome after completion of the entire series of assessments.
Generic Components
This chapter covers the generic components which are relevant to 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 trainees with differing application levels in practice.
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
- Knowledge of the procedure for the 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, strategies applicable to smoking, alcohol, drug abuse, lifestyle changes
- 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
- The need and place for specific types of notes e.g. problem-orientated discharge, letters, concise out-patient reports
- 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
- Ability to enlist patients’ involvement in solving their health problems, providing information, education
- Availing of support provided by voluntary agencies and patient support groups, as well as expert services e.g. detoxification / psychiatric services
- Valuing contributions of health education and disease prevention to health in a community
- Compiling adequate case notes, with 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)
- 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)
- 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
- 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 tool (e.g. ISBAR)

ASSESSMENT & LEARNING METHODS

- ACLS course
- Record of on call experience
- Mini-CEX (acute setting)
- Case based discussions
- Consultant feedback
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
- Disclosure – know 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 according to Data Protection Act 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)

- When and how to report a near miss or adverse event
- Knowledge of preventing and managing near misses and adverse events. 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

- The importance of patient safety relevance in health care setting
- Standardising common processes and procedures – checklists, vigilance
- The multiple factors involved in failures
- Safe healthcare systems and provision of a safe working environment
- The relationship between ‘human factors’ and patient safety
- Safe working practice, role of procedures and protocols in optimal practice
- How to minimise incidence and impact of adverse events
- 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
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
- Ability to learn from errors and near misses to prevent future errors
- Using relevant information from complaints, incident reports, litigation and quality improvement reports in order to control risks
- Minimising errors during invasive procedures by developing and adhering to best-practice guidelines for safe surgery
- Minimising medication errors by practicing safe prescribing principles
- Using the Open Disclosure Process Algorithm
- Managing errors and near-misses
- Managing complaints
- Ethical and legal decision making skills

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
- Patient Safety (on-line) – recommended
- Leadership in Clinical Practice III
- Quality improvement methodology course - recommended
- RCPI Ethics programmes (I-IV)
- Medical Council Guide to Professional Conduct and Ethics
- Reflective learning around ethical dilemmas encountered in clinical practice
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).

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### 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 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 infectious disease requiring notification
- In surgery or during an invasive procedure, understanding the increased risk of infection in these patients 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
- Healthcare Associated Infections (on-line) – recommended
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
- Good Clinical Practice guidelines for seeing and managing patients who are on clinical research trials

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
- Taking a history of drug allergy and 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
- Principles of Antibiotics Use (on-line) – recommended
- Guidance for health and social care providers - Principles of good practice in medication reconciliation (HIQA)
Self-Care and Maintaining Well-Being

Objective:
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 knowledge – understand own psychological strengths and limitations
- Understand how own personality characteristics (such as need for approval, judgemental tendencies, needs for perfection and control) affect relationships with patients and colleagues
- 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 for specific patients
- 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 (love, anger, frustration, vulnerability, intimacy, etc) in “easy” and difficult 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

- Occupational Stress course
- On-going supervision
- Ethics courses
- Leadership in Clinical Practice III
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, knowing how and when to break bad news, how to negotiate cultural, language barriers, dealing with sensory or psychological and/or intellectual impairments, how to deal with challenging or aggressive behaviour
- How to communicate essential information where difficulties exist, how to appropriately utilise the assistance of interpreters, chaperones, and relatives.
- How to deal with anger, 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 concise, problem-orientated statement of facts and opinions (written, verbal or electronic)
- Knowledge of legal context of status of records and reports, of data protection (confidentiality), Freedom of Information (FOI) issues
- Understanding of the relevance to continuity of care and the importance of legible, accessible, records
- Knowing when urgent contact becomes necessary and the appropriate place for verbal, telephone, electronic, 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 to outcome of continuity of care, within and between phases of healthcare management
- The importance of completion of tasks and documentation (e.g. before handover to another team, department, specialty), of identifying outstanding issues and uncertainties
- Knowledge of the required attitudes, skills and behaviours which facilitate continuity of care such as maintaining (legible) records, 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, retain attention avoid 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 risks of information overload
- Interpreting results, significance of findings, diagnosis, explaining objectives, limitations, risks of treatment, using communication adjusted to recipients’ ability to comprehend
- Ability to achieve level of understanding necessary to gain co-operation (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, assistance available
- The importance of obtaining and recording accurate and full information, seeking confirmation from multiple sources
- Knowledge of how to establish facts, identifying issues and responding quickly and appropriately to a complaint received

Skills

- Ability to elicit facts, using a mix of open and closed-ended questions appropriately
- 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 (and attempting to reach) realistic objectives, identifying and prioritising outstanding problems
- Using language, literature (leaflets) diagrams, educational aids and resources appropriately
- Ability to 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
- 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
- Ethics courses
- Leadership in Clinical Practice III
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
- Leadership in Clinical Practice III
- 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

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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

- Leadership in Clinical Practice III
- 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

- Health Research – An Introduction
- Effective Teaching and Supervising Skills course - recommended
- Educational Assessment Skills course - recommended
- Performing audit course
- 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
- Leadership in Clinical Practice III
- Annual audit
- Consultant feedback on management and leadership skills
- Involvement in hospital committees
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, haemochromatosis
- 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, limb girdle, FSH and Emery Dreifuss muscular dystrophies
- 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, hair root extraction 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, including family care workers
- 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 NCMG clinical meeting
- Record pedigree cases
Formal Genetics 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 technologies and their application (including gene therapy)

KNOWLEDGE

- Knowledge of:
  - 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

SKILLS

- Use primary sources of data
- Appreciate 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
- 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

ASSESSMENT & LEARNING METHODS

- Case based Discussion: Review pedigrees
- Case presentations at Grand Rounds
- Study Day - Bayes Theorem, Mutagenesis, calculation of gene frequencies
- Study Day - Ad hoc appropriate genetic courses e.g. mechanisms of origin of numerical and structural chromosome abnormalities
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 of:
  - Patient sessions by genetic counsellor in common genetic conditions
  - Cancer cases
  - Developmental delay cases
  - Cases of common genetic conditions e.g. Cystic Fibrosis, sickle cell anaemia, chromosomal abnormality, an x-linked recessive genetic condition
  - 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 mental retardation (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
- Interpret Variants of Unknown Significance (VOUS)

**ASSESSMENT & LEARNING METHODS**

- Attend paediatric neurology meetings
- Sit in on Huntington's cases
- Sit in on 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 handicap
- 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 London dysmorphology data base

**ASSESSMENT & LEARNING METHODS**

- Present known and unknown cases
- Write up case report
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
- Write up case report
Cancer Genetics

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

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

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 (VOUS)

ASSESSMENT & LEARNING METHODS
- Working with families referred for cancer risk assessment
- Study day: Cancer module
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

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

ASSESSMENT & LEARNING METHODS

- Attendances at neonatal post-mortem examinations,
- Study day – attendance at joint fetal medicine meetings
- Attend fetal medicine unit to observe the following procedures: amniocentesis and chorionic villus sampling and ultrasound scanning of pregnancies.
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.

**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,
- Metabolic tests required in the investigation of developmental delay
- 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
- The principles of gene therapy
- 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 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 (including paediatric & adult & maternal PKU)
- Attendance of adult clinics to include Fabry Renal clinic
- Become familiar with clinical research trials and GCP
- Attend as observer at Newborn Screening 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.
- Cases to include
  - patients with Lysosomal Storage Diseases
  - cases with suspected mitochondrial disease
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)
- 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

**SKILLS**

- Awareness of the importance of informed consent that arise in relation to storage of DNA samples and cell lines
- 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 (e.g. MLINK, Bayesian analysis)
- Be aware of importance of Bioinformatics & be able to do database searches (Decipher & Ensembl)

**ASSESSMENT & LEARNING METHODS**

- Attending chromosome and DNA laboratory
- Case Based discussion: Unusual and molecular chromosome analysis
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
Joint Specialist Clinics

**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
  - 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
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

**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

**ASSESSMENT & LEARNING METHODS**

- Study Day
- Case Based discussion
Minimum Requirements for Training

- These are minimum tracking requirements. This generally means that in practice, trainees will perform above the stated requirements; however, for record tracking purpose, the following figures have been allocated.
- Where the minimum requirement state “1”, there is no allocated minimum – eLogbook will automatically default to “1”

<table>
<thead>
<tr>
<th>Curriculum Requirement</th>
<th>Required/Desirable</th>
<th>Minimum Requirement</th>
<th>Reporting Period</th>
<th>Form Name</th>
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<td>Section 1 - Training Plan</td>
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<td>• Cross City (Paeds) Neurology meeting</td>
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<td>• Paeds Disorders of Sexual Development (Endocrine)</td>
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<td>• Dermatology</td>
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