INSTITUTE
OF MEDICINE

ROYAL COLLEGE OF
PHYSICIANS OF IRELAND

INTERNATIONAL CLINICAL FELLOWSHIP TRAINING IN

Clinical Genetics

This curriculum of training in Clinical Genetics was developed in 2023 by Prof Andrew Greene (Clinical Genetics Consultant) and undergoes revision by 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
1.0	01 July 2023	Keith Farrington	New Curriculum

Table of Contents

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 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 this Subspecialty International Clinical Fellowship Programme (ICFP) is 1 year, following 2/3 years of training in ICFP Gastroenterology.

- Each ICFP is developed by the Royal College of Physicians of Ireland will be specifically designed so as to meet the training needs of participants to support the health service in their home country.
- All 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 of the relevant medical speciality 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.

Core Professional Skills

This section includes the Medical Council guidelines for medical professional conduct, regarding Partnership, Performance and Practice.
These principles are woven within training practice and feedback is formally provided in the Quarterly Evaluations, End of Post, End Year Evaluation.

Partnership

Communication and interpersonal skills

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

Collaboration

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

Health Promotion

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

Caring for patients

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

Performance

Patient safety and ethical practice

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

Organisational behaviour and leadership

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

Wellbeing

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

Practice

Continuing competence and lifelong learning

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

Reflective practice and self-awareness

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

Quality assurance and improvement

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

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
- 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
- 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)
- Analyse and interpret genetic variation from exome and array testing

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

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	Desirable	1	Training Programme	
Cystic Fibrosis	Desirable	1	Training Programme	
General Genetics	Recommended	60	Training Programme	
Cardiac Genetics	Recommended	1	Training Programme	
Cancer Clinic	Desirable	1	Training Programme	
Ward Rounds/Consultations				Clinical Activities
Consultations	Recommended	20	Training Programme	
Procedures/Practical Skills/Surgical Skills				Procedures, Skills, & DOPS
Skin Biopsy	Recommended	5	Training Programme	
Buccal Swabs	Recommended	5	Training Programme	
Additional/Special Experience Gained				Clinical Activities
Newborn screening laboratory	Desirable	1	Training Programme	
Chromosome and DNA laboratory	Recommended	1	Training Programme	
Patient sessions with Genetic counselor	Recommended	1	Training Programme	
Cardiac Genetics	Desirable	1	Training Programme	
Time in fetal unit	Recommended	1	Training Programme	
Huntington's cases	Recommended	1	Training Programme	
Neonatal post-mortem examination	Recommended	1	Training Programme	
Record of cases	Recommended			Cases

Curriculum Requirement	Required/Desirable	Minimum Requirement	Reporting Period	Form Name
X-linked Pedigree cases	Recommended	5	Training Programme	
Cancer cases	Recommended	1	Training Programme	
Development delay cases	Recommended	5	Training Programme	
Cases of common genetic conditions e.g. Cystic Fibrosis, sickle cell anaemia, chromosomal disorder an X-linked recessive genetic condition	Recommended	1	Training Programme	
Adult neurology cases	Recommended	1	Training Programme	
Lysosomal storage diseases (8 patients , 2 with suspected mitochondrial disease)	Recommended	1	Training Programme	
Management Experience	Desirable	1	Training Programme	Management Experience
Section 3 - Educational Activities				
Courses				Teaching Attendance
RCPI Mastering Communications	Desirable	1	Training Programme	
RCPI Wellbeing	Desirable	1	Training Programme	
RCPI Performing Audit	Desirable	1	Training Programme	
RCPI An Introduction to Health Research	Desirable	1	Training Programme	
RCPI HST Leadership in Clinical Practice	Desirable	1	Training Programme	
RCPI Ethics Foundation	Desirable	1	Training Programme	
RCPI Ethics for GIM	Desirable	1	Training Programme	
RCPI Cancer Genetics	Optional			
ACLS	Desirable	1	Training Programme	
ESHG Recognised Online Clinical Bioinformatics course (To be agreed with Trainer)	Recommended	1	Training Programme	
St Georges Online MOOC Genomics Course	Recommended	1	Training Programme	
Manchester Online Bioinformatics Course	Recommended	1	Training Programme	
ACMG curated educational material for Trainees	Recommended	1	Training Programme	
Educational Material about NGS and genome sequencing and examples on the US Clingen site	Recommended	1	Training Programme	
Bertinoro ESHG course on genomics and NGS	Recommended	1	Training Programme	
The Fundamentals of GDPR (HSE Land)	Recommended	1	Training Programme	
Study Days	Recommended	1	Training Programme	Teaching Attendance

Curriculum Requirement	Required/Desirable	Minimum Requirement	Reporting Period	Form Name
National/International meetings (minimum 1 per year)	Recommended	1	Training Programme	Additional Professional Experience
In-house activities				Attendance at Hospital Based Learning
Paediatric neurology meetings	Recommended	1	Training Programme	
Grand Rounds	Recommended	1	Training Programme	
Specialty Meeting – Neurology, endocrinology, dermatology	Recommended	6	Training Programme	
UK Dysmorphology group meetings	Recommended	1	Training Programme	
Journal Club	Recommended	1	Training Programme	
Pathology Conferences	Desirable	1	Training Programme	
MDT Meetings • Paediatric cancer genetics Meeting • Cross City (Paediatrics) Neurology meeting • Paediatric Disorders of Sexual Development (Endocrine) • Fetal Medicine MDT	Recommended	1	Training Programme	
Seminar	Recommended	2	Training Programme	
Lecture	Recommended	2	Training Programme	
Research Activities	Desirable	1	Training Programme	Research Activities
Audit activities	Recommended	2	Training Programme	Audit & QI
Publications	Desirable	1	Training Programme	Additional Professional Experience
Presentations	Recommended	1	Training Programme	Additional Professional Experience
Presentation at genetics meeting	Recommended	2	Training Programme	
Additional Qualifications	Desirable	1	Training Programme	Additional Professional Experience
Section 4 - Assessments				
CBD	Recommended	5	Training Programme	Case Based Discussion
DOPS	Recommended			Procedures, Skills, & DOPS
Skin Biopsy	Recommended	5	Training Programme	

Curriculum Requirement	Required/Desirable	Minimum Requirement	Reporting Period	Form Name
Buccal swab	Recommended	5	Training Programme	
Pedigree drawing	Recommended	10	Training Programme	
Mini-CEX (At least two Mini-CEX assessments)	Recommended	2	Training Programme	Mini-CEX
Quarterly Assessment/End of Post Assessment	Required	4	Year of Training	Quarterly/End of Post Assessment
End of Year Evaluation	Required	1	Year of Training	End of Year Evaluation