POSTGRADUATE INSTITUTE OF MEDICINE
UNIVERSITY OF COLOMBO

PROSPECTUS

BOARD CERTIFICATION IN CLINICAL GENETICS
(To be effective from 2019)

BOARD OF STUDY IN PAEDIATRICS
SPECIALITY BOARD IN CLINICAL GENETICS

BOM Approved – 06.07.2019
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CONTENTS

"This prospectus is made under the provisions of the Universities Act, the Postgraduate Institute of Medicine Ordinance, and the General By-Laws No. 1 of 2016 and By-Laws No. 2 of 2016 for Degree of Doctor of Medicine (MD) and Board Certification as a Specialist"

1. Background

Clinical Genetics is the medical speciality concerned with the diagnosis and medical management of inherited disorders and birth defects, estimation of genetic risks and provision of genetic counselling for family members. The Clinical Geneticists work in close collaboration with other medical specialists and laboratory scientists in a collaborative work environment.

Today Clinical Genetics has become very important in clinical practice. The National Health Strategic Master Plan for 2016 to 2025 of the Ministry of Health, Nutrition and Indigenous Medicine has identified the need for development of Clinical Genetics services as a national programme in Sri Lanka. One of the goals of this programme is to train adequate number of MD qualified Clinical Geneticists to fulfill the national need.

In fulfilling this requirement, training in Clinical Genetics will be offered to those who have demonstrated competence in Adult General Medicine and Paediatrics as evidenced by passing the MD (Medicine) or MD(Paediatrics) examination at the PGIM. This will ensure that the Clinical Geneticist would be competent in delivering total medical care to patients with genetic disorders, the majority of whom would require screening and management of general medical/paediatric conditions and coordination of their care with other medical and surgical specialists.

2. Entry criteria, Selection Process and Intake

2.1. Entry Criteria

A candidate who fulfills the following requirements are eligible to be enrolled into the programme;

a) passed the MD (Medicine) or MD (Paediatrics) examination
b) has not applied or has not already enrolled into any other post-MD training programme at the PGIM
c) has not obtained Board Certification in any field

2.2. Selection Process

The positions available will be offered to the trainees by the Speciality Board in Clinical Genetics on the basis of the order of merit at the relevant MD examination.

2.3. Intake

The number of trainees to be enrolled each year shall be predetermined by the Speciality Board in Clinical Genetics in collaboration with the Board of Study in Medicine and Board of Study in Paediatrics as approved by the Board Board of Management. The available training opportunities will be stated in the exam circulars published for the MD Medicine and MD Paediatrics examinations.

3. Aim and Learning Outcome

3.1. Aim

The aim of the programme is to produce a clinical geneticist who would be able to diagnose and manage genetic disorders and develop further the speciality in clinical genetics and the clinical genetic services in Sri Lanka to the highest international standards.

3.2. Learning Outcomes

At the end of the programme, a Board-Certified Specialist in Clinical Genetics shall be able to;

a. diagnose and manage pathological states presenting in clinical genetics
b. organize and lead genetic services fitting the needs of a particular context and evaluate its outcomes
c. conduct audits and scientific research, with a view to contributing to scientific knowledge in the field and participating in the task of improving genetic services in the community
d. communicate effectively complex concepts and genetic tests results to families enabling them to make informed decisions
e. educate other health professionals, medical and postgraduate students, and the general public on relevant aspects in clinical genetics including prevention and health promotion
f. critically appraise research publications and practice evidence-based medicine
g. maintain highest standards of professionalism, moral and ethical conduct as a clinical geneticist
h. engage in continuous professional development and become a lifelong learner
i. play a critical role in the discourse around ethical and moral aspects related to the application of new genetic developments in healthcare

4. Structure of the Training Programme

The Board Certification in Clinical Genetics programme shall be a three (3) year programme consisting of the following components.

4.1. Local Training

The local training shall be for one and a half years (18 months) at centres in Sri Lanka accredited by the Speciality Board in Clinical Genetics for Clinical Genetics training. The centres shall be located in Teaching Hospitals, Special Units at the Ministry of Health, Faculties

of Medicine and in accredited private sector institutions when the said expertise is not available within the state sector. A trainee shall spend the designated period of time in centres recognized to provide following training:

Area of training	Duration	Training centres*
General Medicine/General Paediatrics*	2 months	National Hospital of Sri Lanka (NHSL) Lady Ridgeway Hospital (LRH)
Paediatric genetics and dysmorphology	3 months	Lady Ridgeway Hospital (LRH)
Neurogenetics	3 months	National Hospital of Sri Lanka (NHSL) North Colombo Teaching Hospital South Colombo Teaching Hospital
Cardiac Genetics	2 months	National Hospital of Sri Lanka (NHSL)
Cancer Genetics	2 months	National Cancer Institute, Maharagama
Prenatal Diagnosis and Fetal Dysmorphology	3 months	Lady Ridgeway Hospital (LRH) Castle Street Hospital for Women De Zoysa Maternity Hospital for Women
Laboratory Genetics (Cytogenetic and Molecular genetic)	3 months	Faculty of Medicine, University of Colombo Pathology laboratories in government institutions Private sector institutions

* The available training centres may change depending on the availability of trainers, number of trainees that can be accommodated at any given time and other facilities available for training. New training centres may also be added following being accredited by the Board.

* Trainees with a general medicine background shall spend eight weeks (2 months) in a Paediatric General Medicine setting while trainees with Paediatrics background shall spend eight weeks (2 months) in an Adult General Medicine setting. Trainees are expected to gain general medicine/paediatric training during this period. Altogether, a trainee with Paediatrics background shall spend two thirds of the training in adult medicine settings while a trainee with a General Medicine background shall spend two thirds of the training in Paediatric settings.

A trainee shall maintain a log of attendance throughout the training and shall obtain the signature of the assigned trainer on completion of each attachment. A trainee must maintain 80% attendance for each attachment and shall submit the log of attendance to the PGIM on completion of the local training component. The complete log shall be part of the portfolio.

4.2. Overseas Training

The post-MD overseas training shall be for a minimum period of one and a half years (18 months). The training centre shall be accredited by the Speciality Board in Clinical Genetics and shall provide the trainee with the opportunity to gain the desirable learning related to Clinical Genetics as designated by the programme.

5. Content areas

The content areas have been described in detail in Annex 1. The curriculum has been developed in-line with the Specialty Training Curriculum for Clinical Genetics of the Joint Royal Colleges of Physicians Training Board, UK and Framework for Development of Physician Competencies in Genomic Medicine: report of the Competencies Working Group of the Inter-Society Coordinating Committee for Physician Education in Genomics, USA. The relevant content areas have been adopted to fit the Sri Lankan context and training needs of a Clinical Geneticist.

6. Teaching and Learning Activities

Trainees will achieve the competencies described in the curriculum through a variety of learning methods. There will be a balance of different modes of learning from formal teaching programmes to experiential learning 'on the job'. The proportion of time allocated to different learning methods may vary depending on the nature of the attachment within a rotation. There are many opportunities for trainees to learn with other Clinical Genetics trainees. There will be study days organised at specific times of the year specifically for trainees from all centres to capitalise on specialist educational input from other centres involved.

6.1. Acquisition of theoretical Knowledge

The acquisition of theoretical knowledge would be facilitated through a variety of methods. These may include;

- Case presentations
- Journal clubs
- Lectures and small group teaching
- Grand Rounds
- Clinical skills demonstrations and teaching
- Journal clubs
- Joint specialty meetings

Trainees are expected to engage in independent self-directed learning. They are expected to use this time in a variety of ways depending upon their stage of learning. Suggested activities include:

- reading, including web-based material and journals
- maintenance of personal portfolio (self-assessment, reflective learning, personal development plan) (Please refer to section 8)
- audit and research projects

Trainees may also utilize external learning opportunities in gaining the relevant knowledge and skills as designated in the curriculum. These opportunities may include;

- attendance at regional and national meetings e.g. Dysmorphology Club, Cancer Genetics Group relevant to the current component of training being undertaken.
- attendance and presentation at national conferences
- attendance and presentation at international conferences
- participation in the work of patient support groups.

Trainees are expected to maintain a reflective log (Please see Annex 2) of each of these activities and include it in their portfolio as explained in section 8.

6.2. Clinical Training

The clinical training placements will be in-service training under direct supervision of a designated trainer approved by the Postgraduate Institute of Medicine. The content of work-based experiential learning is decided by the trainer but includes active participation in:

- Out-patient clinics and ward referrals: After initial induction, trainees will see patients in outpatient clinics and wards in both the training unit as well as in outreach clinics and wards, supervised by the trainer. The degree of responsibility taken by the trainee will increase as competency increases. As experience and clinical competence increase, trainees will assess 'new' and 'review' patients and present their findings to their trainer.

- Multi-disciplinary team meetings: There are many situations where clinical problems are discussed with clinicians in other disciplines. These provide excellent opportunities for observation of and participation in clinical reasoning.

- Co-counselling sessions: Co-counselling with experienced genetic counsellors enables constructive feedback on the counselling sessions and the development of appropriate professional behaviours in dealing with patients and families. The trainee observes and undertakes the whole process of genetic counselling from initial contact to follow-up.

- Laboratory sessions**:** Understanding the methodologies undertaken in the genetics laboratory is essential for the clinical geneticist who is required to request investigations appropriately and explain and interpret the results of tests for patients and their families as well as fellow professionals. The trainees are expected to achieve learning through hands on experience in the laboratories. Each trainee will spend time in both cytogenetic and molecular genetics laboratories learning the basic procedures and methodologies employed. Apart from laboratory bench experience there will be opportunities for small group tutorials with laboratory scientists.

- Specialised Clinic Sessions: Many genetic disorders are rare and short blocks of attendance and observation in Specialised Clinics are incorporated within the training as and when opportunities arise. These opportunities may include;

    - specialist clinics within the Clinical Genetics service
    - multi-disciplinary clinics (joint clinics with other specialists)

- clinics held by non-genetic specialists in areas that impact on clinical genetic practice (e.g. fetal and reproductive medicine)
- clinics held by non-genetic specialists that allow a greater understanding of the clinical management of conditions, both common (e.g. Oncology Clinics) or rare disorders (e.g. Inherited Metabolic Disorders) and the management of genetic disorders in non-genetic settings (e.g. Haematology Clinics)

The trainee must maintain a reflective log of clinical training according to the format in Annex 2 and include the same in the portfolio.

6.3. Research Project

Trainees are expected to carry out a research project during their training. The Research Project should be undertaken at the commencement of training. It should be a clinical study including a laboratory component. The Board will appoint a supervisor for the Research Project at the commencement of the programme.

6.3.1. Submission of the research proposal

A comprehensive research proposal prepared in accordance with the structure and guidelines given in Annex 3 shall be submitted to the Board within three months of commencing the training programme.

The proposal should be prepared under the supervision of the assigned supervisor and be endorsed by the supervisor prior to submission to the Board.

The submitted proposal will be evaluated by the Board and the trainee can expect to know the outcome of this evaluation within one month of submission.

6.3.2. Conduct of the research

Trainees must obtain ethical approval before the commencement of the research.

The candidate may continue to conduct the research project during the foreign training under the supervision of the local supervisor who shall remain the supervisor of the research project for its total duration.

6.3.3. Completion of the research project

According to the General Regulations and Guidelines of the PGIM, the acceptance of the research project by the Board of Study will be based on fulfilment of either of the following:

- Publication of the research findings as an original full paper (not case reports) in a peer reviewed journal (preferably indexed) with the trainee as first author; or

- Submission of a detailed project report to the Board and be successful in its assessment.

The project report or the full paper published in a journal shall be part of the portfolio.

6.4. Clinical Audit

The trainee must carry out a clinical audit of genetic services in a training unit that the trainee is assigned to and present it to the Institution. An example of an audit could be 'An audit of genetic referral patterns.

The trainee must present the findings of the audit at a clinical meeting in the institution where the unit is located or at a scientific session of a university or a professional association.
The manuscript prepared based on the clinical audit shall be part of the portfolio.

6.5. Case reports

The trainee must complete 5 case reports and include the same in the portfolio. The five cases must consist of one case each in Paediatric Genetics and Dysmorphology, Neurogenetics, Cardiac Genetics, Cancer Genetics; and Prenatal Diagnosis and Fetal Dysmorphology.

The trainee may use the following structure when preparing the case reports.

- Abstract: The manuscript should contain an abstract. The abstract should be self-contained and citation-free and should not exceed 200 words.

- Introduction: This section should be succinct, with no subheadings.

- Case Presentation: This section should present all of the relevant information about the patient in the case being reported, as well as a full description of the patient's symptoms, diagnosis, treatment, and outcome.

- Discussion: This section should provide context for the case being reported and provide any necessary explanation of specific treatment decisions.

The length of each case report shall not exceed 1500 words.

7. Progress Monitoring

Following progress monitoring mechanism are incorporated into the training programme.

7.1. Clinical Training Progress Reports

The trainers shall submit the Clinical Training Progress Report to the Board every three months using the form in Annex 4. The progress reports shall continue during both local and overseas training. The trainees should liaise with the trainers and ensure that the progress reports reach the PGIM in time.

The trainers shall evaluate the progress of the trainees in relation to following areas:

1. Theoretical knowledge
2. Clinical skills
3. Clinical judgment
4. Attitudes
    a. reliability
    b. self-motivation
    c. team leadership
    d. teaching commitment
5. Professional and ethical conduct

The trainee should liaise with the trainers and make sure that the reports are received by the PGIM in time (within two weeks of the due date).

Satisfactory progress would be determined by the attainment of above average rating in 75% of indicators. A rating of poor in any indicator would signify unsatisfactory progress irrespective of the rating attained in other indicators.

Suitable and appropriate action will be taken by the Board in accordance with the General Regulations and Guidelines of the PGIM in the event of the receipt of an unsatisfactory or adverse clinical training progress report at any stage of training.

Satisfactory Clinical Training Progress Reports are a mandatory requirement to qualify for the Pre-Board Certification Assessment (PBCA).

7.2. Research Progress Reports

The trainee should submit a progress report of the research project to the Board every six months using the form in Annex 5.

The trainee should liaise with the trainer and make sure that the reports are received by the PGIM in time.

Suitable and appropriate action will be taken by the Board of Study according to the General Regulations and Guidelines of the PGIM in the event of the receipt of an unsatisfactory or adverse research progress report at any stage of training.

Satisfactory Research Progress Reports are a mandatory requirement to qualify for the Pre-Board Certification Assessment.

7.3. Multi Source Feedback

Trainees must complete at least one Multi Source Feedback (MSF) during the training.

The trainee shall follow the guidelines given in the PGIM website when completing MSF and shall incorporate the MSF report provided by the PGIM in relation to each of the MSF in the relevant section of the portfolio.

7.4. Formative Assessments

The trainer shall carry out a formative assessment of the trainee at the end of every six months according to the format given in Annex 6.

Formative assessment reports should be submitted to the PGIM within 2 weeks of the due date.

Suitable and appropriate action will be taken by the Board according to the General Regulations and Guidelines of the PGIM in the event of the receipt of an unsatisfactory or adverse formative assessment report at any stage of training.

Satisfactory Formative Assessment Reports are a mandatory requirement to qualify for the Pre-Board Certification Assessment.

7.5. Knowledge Based Assessment

A trainee shall complete a knowledge based formative assessment conducted by the Board on completion of first and second year of training (at 12 and 24 months).

The assessment shall be formative and would consist of a single paper consisting of 20 compulsory Short Answer Questions (SAQs), to be answered in three hours.

On completion of the assessment, the Board shall appoint a trainer to evaluate the knowledge-based assessment and provide feedback to the trainee. The trainee in consultation with the trainer shall formulate a plan of action that would enable the trainee to overcome the areas that he or she has been found deficient in knowledge.

The two knowledge-based assessments, the feedback received, and the plan of action shall be part of the portfolio.

8. Portfolio

The trainee shall maintain a portfolio throughout the training period in accordance with the given structure. A softcopy of the portfolio shall be submitted to the Board upon requested and at regular six-monthly intervals for formative evaluation and feedback during both local and foreign training.

Trainees are expected to structure the training portfolio using the given headings and the content as follows:

- Subject expertise
    - Reflective logs related to academic activities taken part during training (as indicated in 6.1)
    - Reflective logs related to clinical training activities taken part during training (as indicated in 6.2)

  - Reflective logs pertaining to external events relevant to Clinical Genetics taken part during the training (as indicated in 6.1)
  - Five case reports in manuscript format or if a case has been published, the published manuscript [Please also refer to 6.5]. The trainee should be the first author of any published manuscript
  - Clinical training progress reports
  - Evidence of reading literature around Clinical Genetics including a brief reflection of learning
  - Formative assessments
  - Knowledge-based assessment report

- Teaching
  - Evidence of undertaking teaching activities in areas related to Clinical Genetics for medical students, medical doctors or other health staff.

- Research and audit
  - Published manuscript of the research project [In the case of an accepted manuscript awaiting publication: the manuscript, with the letter/email of acceptance]; or a detailed project report (Please refer to 6.3 for details).
  - The Audit Report with evidence of presentation to a clinical meeting in the Institute (eg. Letter from the Head of the Institute) or a professional association (eg. Published abstract or if the abstract was not published, a letter from the association confirming presentation) (Please also refer to 6.4) [Note: If the audit has been published, the published manuscript. The trainee should be the first author of any published manuscript.].

- Ethics and professionalism
  - Evidence of taking part in the 'Professionalism Strand of the PGIM' including a brief reflection on professionalism in medicine.
  - Multi-Source Feedback reports

- Information Technology
  - Participation in training programmes / workshops related to information technology.
  - Evidence of searching for information and application of findings in practice
  - Evidence for developing online content, presentations and other electronic content related to Clinical Genetics.

- Lifelong Learning
  - Participation in conferences and meetings
  - Membership and contribution to knowledge through academic/ professional organizations in Clinical Genetics in advancing research and practice.
  - Evidence of continued interest in a specific area in Clinical Genetics

The completed portfolio shall be submitted to the PGIM prior to Board Certification and shall be the basis on which the Pre-Board Certification Assessment is conducted. When the trainee

is eligible for PBCA, 3 copies of the completed portfolio should be submitted to the PGIM Examinations Branch.

9. Trainers and Training Units

Local training shall be provided in institutions/units that provide clinical and/or diagnostic genetic and related services accredited for training by the PGIM. Each training unit shall undergo an accreditation process as defined in the General Regulations and Guidelines of the PGIM.

Trainers assigned shall fulfill the general PGIM criteria for being recognized as a trainer.

Overseas training shall be in centres approved by the PGIM under the supervision of trainers specialized in Clinical Genetics as approved the Board.

10. Pre-Board Certification Assessment (PBCA)

10.1. Eligibility

The trainee shall be eligible to appear for a PBCA after having satisfactorily completed the following:

1. Completion of the local training programme with at least 80% attendance at each clinical attachment and satisfactory Clinical Training Progress Reports submitted every three months
2. Completion of the overseas training programme with satisfactory Clinical Training Progress Reports submitted every three months
3. Satisfactory Research Progress Reports submitted every six months
4. Submission and acceptance of at least two MSF
5. Successful completion and acceptance of the research project.
6. Successful completion and acceptance of an audit.
7. Successful completion and acceptance of five case reports.
8. Completion of two knowledge-based assessments at 12 and 24 months of training
9. Successful completion and acceptance of the portfolio

10.2. Format of the PBCA

The PBCA shall comprise of a portfolio assessment. The portfolio assessment shall take the form of a viva voce examination based on the portfolio. The candidate will be asked to make a presentation based on the portfolio for 10 minutes and the examiners shall then question the candidate on the contents of the portfolio for 20 minutes.

The assessment shall be carried out by three (3) independent examiners appointed by the Board and approved by the Senate of the University of Colombo. One of the examiners will be from outside the discipline to improve objectivity.

10.3. Requirements to pass the PBCA

The portfolio shall be marked in accordance with the marking scheme provided in Annex 7. A candidate must obtain a satisfactory grade for all components as described in the marking scheme to pass the PBCA.

10.4. Failed Candidates

If the PBCA results in an unsatisfactory outcome the candidate would not be given immediate Board Certification and the examiners shall provide the candidate with a written feedback on how the portfolio should be improved in order to reach the required standard.

The candidate should re-submit the portfolio within a defined period of time (up to 3 – 6 months), and face another oral examination based on the re-submitted portfolio. If the candidate is successful at this 2nd oral examination, the date of Board Certification shall be backdated. If unsuccessful again, the date of Board Certification will be the date of passing the subsequent PBCA following further training for a minimum period of six months in a unit selected by the Board.

11. Board Certification in Clinical Genetics

11.1. Requirements for Board Certification

In order to be eligible for Board Certification, a trainee is required to pass the PBCA.

11.2. Board Certification

A trainee who has successfully completed the Pre-Board Certification Assessment is eligible for Board Certification as a Specialist in Clinical Genetics, on the recommendation of the Speciality Board in Clinical Genetics.

ANNEX 1 – CLINICAL GENETICS CURRICULUM

(Adapted from the Clinical Genetics Specialty Training curriculum of the Joint Royal Colleges of Physicians Training Board, UK)

Outline

Legend: Research project (RP), Clinical Audit (CA), Case Reports (CR), Clinical Training Progress Reports (CTPR), Research Progress Reports (RPR), Multisource Feedback (MSF), Formative Assessments (FA), Knowledge Based Assessments (KBA), Portfolio (PF)

Area	Content	Programme outcomes	Assessments
A. Good Clinical Care	History, Examination, Investigations, Safe Prescribing, Management & Note keeping Skills	a, f, h	CTPR, FA, PF
	Pre-Clinic Preparation		
	History		
	Examination		
	Investigations Including Imaging		
	Diagnosis and Management		
	Note- Keeping, Letters, etc		
	Time Management and Decision Making		
B. Procedures	Phlebotomy	a	CTPR, FA, PF
	Skin Biopsy		
	Clinical Photography		
C. Communication Skills and Genetic Counselling	Breaking Bad News	d, e, i	CTPR, FA, PF
	Specific Genetic Issues		
	Complaints		
D. Genetics and Basic Sciences	Cellular and molecular mechanisms that underpin inheritance in man	a, e, f	FA, KBA
	Social and ethical implications of genetic knowledge		
	Patterns of inheritance		
	Risk assessment		
	Emerging genetic technologies including gene therapy		

E. Common Genetic Referrals	Specialist diagnosis, assessment and genetic counselling	a, d	FA, PF
F. Neurogenetics	Genetic causes of central and peripheral nervous system dysfunction	a	CR, FA, KBA, PF
G. Paediatric Genetics and Dysmorphology	Syndrome diagnosis in children	a	CR, FA, KBA, PF
H. Cancer Genetics	Diagnose rare cancer syndromes	a	CR, FA, KBA, PF
	Common cancers and single gene basis		
	Targeted screening		
	Molecular genetic testing		
I. Prenatal Diagnosis and Fetal Dysmorphology	Genetic assessment of foetus	a	CR, FA, KBA, PF
	Prognosis and inheritance of genetic disorders.		
	Legal framework		
	Assessment of foetal abnormality during pregnancy		
	Counselling parents		
J. Cardiac Genetics	Inherited cardiac conditions (ICC)	a	CR, FA, KBA, PF
	Targeted screening		
	Molecular genetic testing		
K. Laboratory Genetics	Genetic laboratory results in a clinical setting	a	FA, KBA, PF
L. Genomic Medicine	Genomic Testing	a, d	FA, KBA, PF
	Patient Treatment Based on Genomic Results		
	Somatic Genomics		
	Microbial and Genomic Information		
M. Maintaining Trust	Professional Behaviour	g, i	MSF, CTPR, FA
	Continuity of Care		
	Doctor-Patient Relationship		
	Recognising Own Limitations		
	Stress & Personal Health		
	Life-Long Learning		
N. Ethics and Legal Issues	Principles of Medical Ethics and Confidentiality	g, i	CTPR, FA, PF
	Informed Consent		

	Legal Framework for Practice		
O. Organisation and Provision Of Genetics Services For Populations	Genetic registers Legal and ethical issues	a, b, d, e	CTPR, FA, PF
P. Patient Education and Disease Prevention	Educating Patients About Disease, Investigations and Management	a, e	PF
	Managing Long-Term Conditions and Promoting Patient Self-Care		
Q. Working with Colleagues	Interactions Between:	b, d, e	MSF
	Hospital & General Practice		
	Hospital & Other Agencies e.g. Social Services		
	Medical and Surgical Specialties		
R. Teaching and Educational Supervision	Principles of teaching, learning and assessments and its application in genetics teaching.	e, h	CTPR, FA, PF
S. Research	Plan and analyse research	c	RP, RPR, PF
T. Clinical Governance	Evidence-Based Medicine	c, f	RP, FA, PF
	Audit		
	Patient Safety		
U. Structure of The Health Services And The Principles Of Management	Structure of the Health Services and Principles of Management	b, g	KBA, PF
V. Information Technology, Computer Assisted Learning and Information Management	Information Technology for Patient Care and own Personal Development Management of health information.	a, c, d, e, h	PF, FA

Note : Please refer to the detailed curriculum below for further details regarding the content areas and learning outcomes. The assessment methods mentioned pertaining to learning outcomes/content may change as per the decisions made by the Board.

Detailed curriculum:

A. **GOOD CLINICAL CARE**

History, Examination, Investigations, Safe Prescribing, Management & Note keeping Skills:

Pre-Clinic Preparation

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

- Knowledge of relevant disorder acquired by background reading

Skills

- Be able to review medical records and identify information sources including databases and literature searches

Behaviours

- Appreciate the importance of identifying key issues and being prepared to deal with these

History

Knowledge

- Define the patterns of symptoms found in patients presenting with genetic disease
- Recognise reliable and unreliable family history data and identify sources for verification
- Recognises importance of different elements of history
- Recognises that patients do not present history in structured fashion
- Knows likely causes and risk factors for conditions relevant to mode of presentation
- Recognise that the patient's agenda and the history should inform examination, investigation and management

Skills

- Be able to take and analyse a clinical history in a relevant, succinct and logical manner
- Be able to overcome difficulties of language, physical and mental impairment
- Use interpreters and advocates appropriately
- Elicit family history information in a sensitive and understanding manner
- Draw complex pedigrees accurately, including consanguinity loops, recording appropriate information
- Manages time and draws consultation to a close appropriately
- Recognises that effective history taking in non-urgent cases may require several discussions with the patient and other parties, over time
- Supplements history with standardised instruments or questionnaires when relevant
- Manages alternative and conflicting views from family, carers, friends and members of the multi-professional team
- Assimilates history from the available information from patient and other sources including members of the multi-professional team

- Recognises and interprets appropriately the use of nonverbal communication from patients and carers
- Focuses on relevant aspects of history
- Maintains focus despite multiple and often conflicting agendas

Behaviours

- Show empathy with patients and other family members
- Appreciate the importance of psychological and social factors of patients and relatives in genetic disease
- 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
- Shows respect and behaves in accordance with Good Medical Practice

<u>Examination</u>

Knowledge

- Define the pathophysiological basis of physical signs
- Define the clinical signs found in genetic diseases
- Understands the need for a targeted and relevant clinical examination
- Understands the basis for clinical signs and the relevance of positive and negative physical signs
- Recognises constraints to performing physical examination and strategies that may be used to overcome them
- Recognises the limitations of physical examination and the need for adjunctive forms of assessment to confirm diagnosis
- Recognise when the offer/ use of a chaperone is appropriate or required

Skills

- Be able to perform a reliable and appropriate examination to elicit relevant signs of genetic disease
- Perform examination appropriately in situations involving cultural sensitivity
- Understand when additional specialist examination is required
- Performs an examination relevant to the presentation and risk factors that is valid, targeted and time efficient
- Recognises the possibility of deliberate harm (both self-harm and harm by others) in vulnerable patients and report to appropriate agencies

Behaviours

- Respect patients' dignity and confidentiality
- Appropriately involve relatives
- In particular ensure examination whilst clinically appropriate considers social, cultural and religious boundaries to examination, appropriately communicates and makes alternative arrangements where necessary

Investigations Including Imaging

Knowledge
- Know the predictive value of results of investigations
- Define the pathophysiological basis of investigations
- Define the indications for investigations
- Define the risks and benefits of investigations
- Know the cost effectiveness of individual investigation

Skills
- Ability to prioritise investigations and interpret the results
- Ability to perform investigations competently where relevant
- Ability to liaise and discuss investigations with colleagues and to order them appropriately

Behaviours
- Willingness to explain to patient and where necessary family the rationale for investigations, and possible unwanted effects

Diagnosis and Management

Knowledge
- Recognise pitfalls in single gene inheritance including variable expressivity and reduced penetrance, somatic and gonadal mosaicism
- Be able to formulate differential diagnoses for genetic disorders

Skills
- Present genetic information to a patient in a sensitive and understanding manner
- Calculate genetic risk in single gene disorders by hand
- Calculate genetic risk by use of a computer programme
- Use computerized genetic databases and registers for information retrieval
- Present undiagnosed cases to colleagues, including dysmorphology club meetings
- Clearly and openly explain management options

Behaviours
- Show appropriate attitudes towards patients and their symptoms and be conscious of religious or other philosophical contexts particularly in respect to prenatal diagnosis
- Sensitivity in breaking bad news
- Appreciate the impact of diagnosing serious genetic conditions on family relationships

Note- Keeping, Letters, etc

Knowledge
- Define the structure, function and legal implications of medical records & medico-legal reports
- Know the relevance of the data protection pertaining to patient confidentiality

Skills

- Record concisely, accurately, confidentially and legibly the appropriate elements of the history, examination, results of investigations, differential diagnosis and management plan

Behaviours

- Timely and cost-effective dictation and communication with medical secretaries
- Prompt and accurate communication with primary care and other agencies
- Show courtesy towards other healthcare professionals

Time Management and Decision Making

Time Management

To demonstrate that the trainee has the knowledge, skills and attitudes to manage time and problems effectively.

Knowledge

- Understand the need to prioritise work according to urgency and importance
- Maintains focus on individual patient needs whilst balancing multiple competing pressures
- Understand the roles, competences and capabilities of other professionals and support workers

Skills

- Recognise when he/she is falling behind and re-prioritise or ask for help
- Organise and manage workload effectively and flexibly
- Make appropriate use of other professionals and support workers
- Employs techniques for improving time management

Behaviours

- Have realistic expectations of tasks to be completed by self and others, particularly patients and their families
- Willingness to consult and work as part of a team
- Identify clinical and clerical tasks requiring attention or predicted to arise

Decision Making

Knowledge

- Define the steps of diagnostic reasoning
- Interpret history and clinical signs
- Conceptualise clinical problem in a medical, psychological and familial context
- Recognise how to use expert advice, clinical guidelines and algorithms
- Recognise and appropriately respond to sources of information accessed by patients
- Recognise the need to determine the most effective or “least bad” treatment both for the individual patient and for a patient cohort
- Define the concepts of disease natural history and assessment of risk
- Describe commonly used statistical methodology

- Know how relative and absolute risks are derived and the meaning of the terms predictive value, sensitivity and specificity in relation to diagnostic tests

Skills

- Interpret clinical features, their reliability and relevance to clinical scenarios including recognition of the breadth of presentation of common disorders
- Incorporates an understanding of the psychological and social elements of clinical scenarios into decision making
- Construct a concise and applicable problem list using available information
- Construct an appropriate management plan in conjunction with the patient, carers and other members of the clinical team and communicate this effectively to the patient, parents and carers securing their agreement to the course of action
- Define the relevance of an estimated risk of a future event to an individual patient
- Use risk calculators appropriately
- Apply quantitative data of risks and benefits of screening and therapeutic intervention to an individual patient
- Search and comprehend medical literature to guide reasoning
- Generate hypothesis within context of clinical likelihood
- Test, refine and verify hypotheses
- Develop problem list and action plan

Behaviours

- Show willingness to discuss intelligibly with a patient the notion and difficulties of prediction of future events, and benefit/risk balance of therapeutic intervention
- Show willingness to adapt and adjust approaches according to the beliefs and preferences of the patient and/or carers
- Show willingness to search for evidence to support clinical decision making
- Demonstrate ability to identify one's own biases and inconsistencies in clinical reasoning

B. PROCEDURES

To demonstrate proficiency in clinical procedures related to genetics.

<u>Phlebotomy</u>

Knowledge

- Knowledge of technique

Skills

- Ability to take blood samples from adults and children, including those with special needs

Behaviours

- Understand the stress of the technique and obtain consent

Skin Biopsy

Knowledge
- Knowledge of technique and indications for use

Skills
- Demonstrate ability to obtain samples suitable for analysis

Behaviours
- Explain procedure appropriately and obtain consent

Clinical Photography

Knowledge
- Knowledge of technique
- Understand importance and confidentiality of photographic records

Skills
- Demonstrate ability to take photographs of sufficient quality for clinical use

- Use of digital photography and storage of data

Behaviours
- Explain the need for clinical photography and obtain consent

C. COMMUNICATION SKILLS AND GENETIC COUNSELLING

Within a Consultation

Acquire and demonstrate effective communication with patients, relatives and colleagues along with the habit of reflection on personal genetic counselling style and effectiveness. ("counselling" in this context means the transmission of information about genetic disease, risk and reproductive options).

Knowledge
- How to structure a consultation appropriately
- The importance of the patient's background, culture, education and preconceptions (beliefs, ideas, concerns, expectations) to the process
- Be aware of social and cultural issues and practices such as:
  - The impact of cultural beliefs and practices on health outcomes
  - Health determinants that affect patients and communities
  - effects of social and cultural issues on access to healthcare, including an understanding of health
  - issues of migrants and refugees
- Specific techniques and methods that facilitate effective and empathic communication
- Understand the importance of the developmental stage when communicating with adolescents and young adults

Skills
- Be able to communicate effectively, both verbally and in writing to patients whose first language may not be English in a manner that they understand

- Give clear information and feedback to patients and share information with relatives when appropriate
- Establish a rapport with the patient and relevant others
- Listen actively and question sensitively to guide the patient and to clarify information in particular with regard to matters that they may find it difficult to discuss, e.g. domestic violence or other abuse
- Utilise open and closed questioning appropriately
- Listen actively and question sensitively to guide the patient and to clarify information
- Identify and manage communication barriers, tailoring language to the individual patient and others and using interpreters when indicated
- Deliver information compassionately, being alert to and managing their and your emotional response (anxiety, antipathy etc.)
- Use, and refer patients to, appropriate written and other evidence based information sources
- Check the patient's/carer's understanding, ensuring that all their concerns/questions have been covered
- Indicate when the consultation nearing its end and conclude with a summary and appropriate action plan; ask the patient to summarise back to check his/her understanding
- Make accurate contemporaneous records of the discussion
- Manage follow-up effectively and safely utilising a variety of methods (e.g. phone call, email, letter)
- Ensure appropriate referral and communications with other healthcare professional resulting from the consultation are made accurately and in a timely manner
- Respect diversity and recognise the benefits it may bring, as well as associated stigma

Behaviours

- Approach the situation with courtesy, empathy, compassion and professionalism, especially by appropriate body language and endeavouring to ensure an appropriate physical environment
- Ensure that the approach is inclusive, and patient centred and respect the diversity of values in patients, carers and colleagues
- Be willing to provide patients with a second opinion
- Accept uncertainty and use different methods of ethical reasoning to come to a balanced decision where complex and conflicting issues are involved
- Demonstrate:
    - Recognising good advice and continuously promoting values based non-prejudicial practice
    - Using authority appropriately and assertively; willing to follow when necessary

<u>Breaking Bad News</u>

Knowledge

- Know how to structure the interview and where it should take place

- Be aware of the normal bereavement process and behaviour
- How bad news is delivered irretrievably affects the subsequent relationship with the patient
- Every patient may desire different levels of explanation and have different responses to bad news
- That bad news is confidential, but the patient may wish to be accompanied
- Breaking bad news can be extremely stressful for the doctor or professional involved
- "Bad news" may be expected or unexpected and it cannot always be predicted
- Sensitive communication of bad news is an essential part of professional practice
- "Bad news" has different connotations depending on the context, individual, social and cultural circumstances

Skills

- Be able to break bad news in steps appropriate to the understanding of the individual and be able to support distress
- Demonstrate to others good practice in breaking bad news
- Recognises the impact of the bad news on the patient, carer,
- supporters, staff members and self
- Encourage questioning and ensure comprehension
- Respond to verbal and visual cues from patients and relatives
- Act with empathy, honesty and sensitivity avoiding undue optimism or pessimism
- Structures the interview inappropriately

Behaviours

- Show empathy, honesty and sensitivity
- Show leadership in breaking bad news
- Respect the different ways people react to bad news
- Ensure appropriate recognition and management of the impact of breaking bad news on the doctor

<u>Specific Genetic Issues</u>

Knowledge

- Knowledge of ethnic difference in the incidence of genetic disease
- Understanding of cross-cultural issues including consanguinity and arranged marriages
- Understanding of religious beliefs and attitudes to prenatal diagnosis and assisted reproduction techniques

Skills

- Use of "non-directive" counselling skills
- Effective use of co-counsellors
- Communication of genetic information and risk to children and adolescents
- Communication with adults and children with learning disability
- Recognising which patients will benefit from referral on to psychological services

Behaviours

- Appreciate patient and family anxieties, both rational and irrational
- Appreciate that every person is influenced by their own culture, ethnicity and beliefs
- Appreciate the importance of genetic counsellors
- Cultivate habit of reflection and discussion with colleagues after counselling sessions
- Readiness to alter practice in light of experience and feed-back

Complaints

Knowledge

- Be aware of the local complaint's procedures
- Be aware of systems of independent review
- Recognise factors likely to lead to complaints (poor communication, dishonesty, clinical errors, adverse clinical outcomes etc.)
- Recognise the impact of complaints and medical error on staff, patients, and the National Health Service

Skills

- Manage dissatisfied patients / relatives
- Contribute to processes whereby complaints are reviewed and learned from
- Explain comprehensibly to the patient the events leading up to a medical error or serious untoward incident, and sources of support for patients and their relatives
- Deliver an appropriate apology and explanation (either of error of for process of investigation of potential error and reporting of the same)
- Distinguish between system and individual errors (personal and organisational)
- Show an ability to learn from previous error
- Recognise when something has gone wrong and identify appropriate staff to communicate this with

Behaviours

- Act with honesty and sensitivity and promptly
- Be prepared to accept responsibility
- Take leadership over complaint issues
- Recognise the impact of complaints and medical error on staff, patients, and the National Health Service
- Contribute to a fair and transparent culture around complaints and errors
- Adopt behaviour likely to prevent causes for complaints

D. FORMAL GENETICS AND BASIC SCIENCES

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

- The chromosomal basis of heredity (mitosis and meiosis)
- Mechanisms of origin of numerical and structural chromosome
- Behaviour of structural chromosome abnormalities at meiosis
- The chemical structure of DNA and replication
- Central dogma of cell biology: transcription and translation.
- 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
- History of genetics

Skills

- 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 – understand the implications of the Hardy-Weinberg equilibrium
- Apply knowledge to interpret results of chromosome and molecular genetic analysis

Behaviours

- Commitment to lifelong self-directed learning
- Appreciation the impact of genetic disorders on individuals and families
- Appreciate potential benefits and harm of new genetic technologies
- Appreciate public concerns about the application of new genetic technologies

E. COMMON GENETIC REFERRALS

To provide the trainee with the skills and knowledge to be able to carry out specialist diagnosis, assessment and genetic counselling genetic conditions

Knowledge

- The genetic basis and clinical features of common genetic conditions
- The medical and surgical complications of common genetic conditions and indications for referral for specialist opinion
- Molecular/cytogenetic testing that is available 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 trial

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
- 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 integrated care pathways and management plans with individuals/families
- Verify diagnoses from old hospital records

Behaviours

- Value the contribution and role of other specialists
- Appreciate role of patient education and support groups e.g. in type 1neurofibromatosis
- Appreciate the role of the general practitioner in management of chronic disease
- Apply good clinical care and counselling skills 1

F. NEUROGENETICS

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
- 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

Skills

- Recognise family history data that suggest familial neurological disease
- Be able to confirm clinical signs in affected individuals
- 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

Behaviours

- Appreciation of family stress caused by risk or eventuality of neurodegeneration
- Appreciate social problems encountered by adults with mild/moderate learning disability
- Appreciate issues involved in predictive testing

G. PAEDIATRIC GENETICS AND DYSMORPHOLOGY

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

Knowledge

- Identify normal developmental milestones and diagnose delayed
- development
- Explain morphogenesis in terms of deformation, malformation, disruption and dysplasia
- Have knowledge of common and rarer dysmorphic syndromes

Skills

- Be able to take a relevant history, and perform an appropriate examination, obtain illustrative photographs
- Have a rational approach to investigation of children with delayed development and/or dysmorphic syndromes.
- Formulate differential diagnoses of unknown syndromes
- Utilise journals and databases used in syndrome identification
- 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
- Present and discuss cases with colleagues

Behaviours

- 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
- Recognise and explain to families when diagnostic work crosses the boundary into research and the constraints that this imposes

H. CANCER GENETICS

Ability to diagnose rare cancer syndromes and recognise when common cancers are likely to have a single gene basis
Ability to recommend targeted screening in individuals who are identified as having increased risk
Coordination of appropriate molecular genetic testing

Knowledge

- The genetic and environmental factors that affect risk of developing cancer
- Current recommendations concerning tumour surveillance in cancer-prone families
- Knowledge of clinical features of genetic cancer syndromes

- Genetic mechanisms in neoplasia: Knudson's two-hit hypothesis
- Knowledge of molecular basis of cancer genetic syndromes
- Knowledge of how inherited and environmental predisposition may affect cancer treatment

Skills

- Be able to take a relevant history, perform an appropriate examination and undertake risk estimation using a variety of methods
- Use of cancer registers and other sources to verify diagnoses
- Use disease registers to support follow-up of affected and at-risk patients
- Assessment of screening protocols for at-risk relatives
- Identify at-risk patients and relatives who are eligible to participate in trials of cancer prevention strategies

Behaviours

- 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
- Understand the impact of cancer risk on individuals and families

I. PRENATAL DIAGNOSIS AND FETAL DYSMORPHOLOGY

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

To acquire the skills and knowledge to assess the risk of potential genetic problems in the foetus prior to pregnancy and to advise parents about the options and procedures open to them within the current legal framework

To develop the skills and knowledge to assess foetal abnormality during pregnancy, to provide parents with information about prognosis, genetic investigations, including post-mortem examination and storage of foetal tissue

Knowledge

- Understand the natural history of prenatally diagnosed conditions, including common single gene and chromosome abnormalities
- Know the indications for and methods of preimplantation and prenatal diagnosis
- Be informed of the latest advances in prenatal diagnosis such as testing free foetal DNA in maternal blood and the potential for non- invasive prenatal DNA diagnosis
- Knowledge of the law pertaining to termination of pregnancy for foetal abnormality
- Know the indications for, process and limitations of foetal post
- mortem examination and issues of consent

- Have knowledge of RCPath guidelines on retention and storage of foetal tissues and the Human Tissues Act

Skills

- Interpret family history data
- Provide genetic advice for women who may undergo preimplantation or prenatal diagnosis
- Formulate differential diagnoses and assess prognosis in collaboration with the foetal medicine team
- Assess risk to foetus when pregnancies are exposed to hazards such as congenital infections, alcohol, ionising irradiation or drugs
- Assess clinical significance of chromosome, DNA and foetal imaging in the context of foetal abnormality
- Evaluate foetal post-mortem findings

Behaviours

- Appreciate the advantages and disadvantages of preimplantation and prenatal diagnosis in each situation
- Non-judgmental appreciation of the ethical and religious dimensions to preimplantation and prenatal diagnosis
- Awareness of the adverse psychological effects of termination of pregnancy for fetal abnormality
- Appreciate the role of relevant patient support groups and other counselling services

J. CARDIAC GENETICS

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

Knowledge

- Knowledge of clinical features of ICC syndromes, including Marfan syndrome and related disorders
- Knowledge of molecular basis of ICC syndromes
- Current recommendations concerning cardiac surveillance in ICC families
- Knowledge of genetic causes of sudden adult death

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 and predictive genetic testing in ICC families
- Identify at-risk patients and relatives who are eligible to participate in prevention strategies (e.g. therapeutic trials)

Behaviours

- Demonstrate awareness of the roles of primary care, specialist nurses and genetic counsellors play 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
- Work as a member of a multidisciplinary team
- Understand the impact of ICC risk on individuals and families; and demonstrate awareness of psychological impact of sudden adult death

K GENOMIC MEDICINE

Genomic Testing

Use genomic testing to guide patient management.

Knowledge

- Discuss the indications for genomic testing—specically the bene ts, risks, and alternatives;
- Explain the implications of placing genomic test results in the patient's medical record;
- Discuss the possibility of incidental findings and how they will be handled;
- Discuss risks of having genomic testing done, e.g., psy- chological implications for the individual as well as the family, the potential for discrimination, and the potential e ect on insurance coverage;
- Explain to the patient issues of costs and nancial cover- age of genomic testing;
- Order, interpret, and communicate the results of appro- priate genomic tests, within the physician's scope of practice;
- Provide referral to an appropriate specialist for genomic testing of a condition outside the physician's scope of practice; and
- Respond to the results of an abnormal genetic screening test, such as newborn screening, including immediate management and appropriate referral.
- Describe the major forms of genomic variability;
- Explain how different genomic changes may result in different phenotypes;
- Recognize that genomic tests require interpretation with respect to the patient's clinical status (e.g., pathogenic, likely pathogenic, and benign);
- Explain the concepts of analytic validity, clinical validity, and clinical utility as they relate to genomic testing; and
- Recognize that medically "nonactionable" genomic results can be useful to the patient and family (i.e., personal utility).

Patient Treatment Based on Genomic Results

Use genomic information to make treatment decisions.

Knowledge

- Identify medical conditions and drug responses that have a strong genetic component;
- Recognize that variants affecting drug responses found in a patient may also have implications for other family members; and
- Discern the potential clinical impact of genetic variation on risk strati cation and individualized treatment.
- Appreciate the importance of genetic diversity in humans and the abundance of genetic variants in each individual genome;
- Identify single-gene disorders that may be amenable to targeted pharmacological therapy;
- Recognize that genomic test results may guide choice of therapy for multifactorial disorders;
- Recognize that there is variability in the phenotypic expression of genetic variants and in response to therapy; and
- Recognize that the effects of some medications are strongly influenced by inherited or somatically acquired genetic variation.

Skills

- Use evidence-based recommendations of professional organizations and others in implementing knowledge gained from genetic discoveries to improve therapeutics;
- Document and periodically reassess therapeutic decision making in the medical record of patients; and
- Incorporate a realistic assessment of personal genomic knowledge and skill into the selection and use of consultants and improve competencies in the wake of these interactions.
- “Treat the patient who has the disease,” i.e., be aware of the patient’s needs as an individual who also has a genetic disease or pharmacogenomic variation.

Behaviours

- Discuss benefits, risks, and alternatives of various preventive and therapeutic approaches driven by genomic findings;
- Communicate clearly with other medical professionals involved in the care of the patient about the therapeutic implications of the genetic information garnered about the patient; and
- Discuss pharmacogenomics implications for future health.
- Respect and guard the privacy of the patient and the family members.
- Maintain the medical knowledge and clinical competence in genomics required for the provision of therapy; and
- Be familiar with the available databases and resources relevant to genetic variation, including ongoing clinical trials involving patients with genetic disorders, pharmacogenomics, and patient-oriented Internet resources from reliable organizations.

- Recognize potential involvement of multiple organ systems in genetic disorders and therefore appreciate the need to seek appropriate consultation with experts in the field; and
- Make medical and genetic information available to other health-care professionals, upon obtaining proper con- sent, while keeping the patient's interests as the primary priority.

Somatic Genomics

Use genomic information to guide the diagnosis and management of cancer and other disorders involving somatic genetic changes.

Knowledge
- Identify or facilitate identification of patients who may bene t from genomic testing of tissue;
- Explain the bene ts and limitations of somatic genomic testing to the patient, including implications regarding treatment of the condition and clarification of his/her prognosis;
- Ensure that tissue biopsy procedures are coordinated to make certain that appropriate and sufficient material is obtained for testing; and
- Integrate genomic testing results into the patient-care plan.
- Explain the concept of somatic genetic change;
- Describe the role of genomic changes in the pathophysiology and treatment of cancer; and
- Explain how genomic testing can be used to guide
    - choice of therapy and adjust drug dosage in patients with cancer.
    - the clinical presentation, suspected pathogen type, and
    - testing method; and
    - Interpret genomics-based tests for diagnosis, monitoring, and treatment of infectious disease.

Skills
- Explain the core strategies for genomic testing for microbial disease;
- Describe how DNA or RNA sequence variations in the microbiome may predict response to therapy and clinical outcomes;
- Explain the potential reasons for false-positive and false- negative microbial genomics-based tests; and
- Explain the importance of "normal" microbiome to health and disease.
- Monitor ongoing testing results and their implications for treatment and prognosis in chronic infection;
- Be aware of new genomic testing methods and their clinical applications and apply when appropriate; and
- Maintain awareness of patterns of infection in your patient population and use genomic tests appropriate to these patterns.
- Work with other health-care professionals to apply infec- tion-control measures when appropriate in both inpatient and outpatient settings; and

- Reassure patients and health-care workers in those situa- tions in which "infection control" is not indicated.

Behaviours
- Explain the results of microbial genomic testing to patients; and
- Explain to patients and families results that signal a risk for contagion and take appropriate containment steps.
- Provide guidance to patients on how to avoid transmission of microbial agents in the community; and
- Appreciate the importance of genomic tests for public health and responsibilities of primary-care physicians in reporting results to the appropriate public health authorities.
- Maintain an awareness of and follow evidence-based guidelines and other professional resources regarding somatic genetic disorders appropriate to the physician's scope of practice.
- Communicate to the patient the importance of genomic testing of his/her tissue sample, including potential impli- cations for treatment and prognosis, and the limitations of genomic testing;
- Address any concerns the patient may have about test results;
- Ensure that specialists involved in a patient's care are com- municating with one another and with the patient; and
- Communicate to patients potential implications for his/ her family.
- Ensure that the patient is aware of what will happen with any tissue samples obtained.
- Maintain a dialogue with the clinical laboratory to ensure that the appropriate test(s) are ordered and interpreted in the context of the patient's clinical status; and
- Be prepared to refer patients to clinical trials designed to evaluate new approaches to cancer therapy.
- Make appropriate referrals to specialists and other health providers and support the patient in ongoing care.
- Keep up to date with progress in the diagnosis and treatment of cancer and other tissue-based disorders.

<u>Microbial and Genomic Information</u>

Use genomic tests that identify microbial contributors to human health and disease, as well as genomic tests that guide therapeutics in infectious diseases.

Knowledge
- Use genomics-based tests for infectious disease instead of classic strategies where appropriate (e.g., based on clinical validity and turnaround time);
- Appreciate the sensitivity and speci city of genomics- based tests for diagnosis of infectious disease based on the clinical presentation, suspected pathogen type, and
- testing method; and

- Interpret genomics-based tests for diagnosis, monitoring, and treatment of infectious disease.

Skills
- Explain the core strategies for genomic testing for micro- bial disease;
- Describe how DNA or RNA sequence variations in the microbiome may predict response to therapy and clinical outcomes;
- Explain the potential reasons for false-positive and false- negative microbial genomics-based tests; and
- Explain the importance of "normal" microbiome to health and disease.
- Practice-based learning and improvement
- Monitor ongoing testing results and their implications for treatment and prognosis in chronic infection;
- Be aware of new genomic testing methods and their clini- cal applications and apply when appropriate; and
- Maintain awareness of patterns of infection in your patient population and use genomic tests appropriate to these patterns.

Behaviours
- Explain the results of microbial genomic testing to patients; and
- Explain to patients and families results that signal a risk for contagion and take appropriate containment steps.
- Provide guidance to patients on how to avoid transmis- sion of microbial agents in the community; and
- Appreciate the importance of genomic tests for public health and responsibilities of primary-care physicians in reporting results to the appropriate public health authorities.
- Work with other health-care professionals to apply infec- tion-control measures when appropriate in both inpatient and outpatient settings; and
- Reassure patients and health-care workers in those situa- tions in which "infection control" is not indicated.
- Identify appropriate specialists and public health o cials who need to be included in the care of the patient with infectious disease and function as a member of the careteam;
- Maintain a dialogue with the clinical laboratory to ensure that the appropriate test(s) are ordered and interpreted in the context of the patient's clinical status; and
- Consult with infectious disease specialists as needed (e.g., to manage unusual or unexpected infection or infection that is highly resistant to treatment).

L. LABORATORY GENETICS

To acquire skills and knowledge to interpret genetic laboratory results within a clinical setting, by completing an attachment in the genetic laboratories

Knowledge

- Understand techniques for conventional cytogenetic analysis in different tissues
- Interpret clinical consequences of chromosome rearrangements
- Understand the principles of FISH analysis and its applications
- Apply array-CGH in different clinical settings and interpret of CNV's (including use of databases such as DECIPHER and ECARUCA)
- Use ISCN nomenclature correctly
- Know the molecular genetic techniques in common usage: DNA extraction, Southern blotting, PCR, MLPA and Sanger sequencing
- Understand the principles and application of next generation sequencing (NGS) technologies including targeted panels, clinical exome sequencing, whole exome sequencing, whole genome sequencing
- Interpret the large data set created from NGS using basic bioinformatics, filtering techniques, clinical and functional data
- Know OMIC technologies and their current and future applications
- Be aware of the Human Genome Variation (HGVS) nomenclature for single gene variants
- Understand the sensitivity and specificity of laboratory tests
- Investigate inborn errors of metabolism through liaison with metabolic disease colleagues and the genetic laboratory
- Be aware of the operation of local and national antenatal and newborn genetic disease screening programmes

Skills

- Undertake genetic risk calculation based on laboratory test results
- (incorporation of genetic test results into Bayesian calculations)
- Interpret results of cytogenetic, molecular cytogenetic, molecular genetics and biochemical tests
- Use databases including ENSEMBL, USCS and locus-specific databases for interpretation of results
- Liaise with laboratory scientists and bioinformaticians in the analysis of test results
- Provide advice to genetic laboratory colleagues on the wording of reports to referring clinicians

Behaviours

- Develop awareness of the importance of informed consent in relation to storage of DNA and cell lines
- Be able to take informed consent when undertaking genomic analyses
- Demonstrate awareness of the potential for incidental findings in genomic analyses and the complexity of these from the patient
- perspective
- Recognise the importance and impact of genetic test results for families and communicate implications of results clearly to them
- Show willingness to liaise with colleagues to interpret laboratory results
- Be able to adapt to new techniques and tests as they arise and incorporate them into clinical practice appropriately

M. MAINTAINING TRUST

Professional Behaviour

Continuity of Care

To ensure that the trainee has the knowledge, skills and attitudes to act in a professional manner at all times.

Knowledge

- Understand the relevance of continuity of care
- Know main methods of ethical reasoning
- Define the concept of modern medical professionalism
- Outline the relevance of professional bodies (SLMC, SLMA, specialist societies, medical defence societies)

Skills

- Make adequate arrangements to cover leave.
- Practise with professionalism including integrity, compassion,
- altruism, continuous improvement, aspiration to excellence, respect of cultural and ethnic diversity, and with regard to the principles of equity
- Work in partnership with patients and members of the widerhealthcare team
- Liaise with colleagues to plan and implement work rotas
- Promote awareness of the doctor's role in utilising healthcare resources optimally and within defined resource constraints
- Recognise and respond appropriately to unprofessional behaviour
- Be able to handle enquiries from the press and other media effectively
- Eliminate discrimination against patients from diverse backgrounds including age, gender, race, culture, disability and sexuality

Behaviours

- Recognise the importance of punctuality and attention to detail
- Recognise personal beliefs and biases and understand their impact on the delivery of health services
- Show willingness to act as a leader, mentor, educator and role model
- Be willing to accept mentoring as a positive contribution to promote personal professional development
- Participate in professional regulation and professional development
- Respect the rights of children, elderly, people with physical, mental, learning or communication difficulties
- Behave with honesty, probity and sensitivity in a non-confrontational manner

Doctor-Patient Relationship

Knowledge

- Understand all aspects of a professional relationship.
- Establish the limiting boundaries surrounding the consultation.

- Outline health needs of particular populations e.g. ethnic minorities and recognise the impact of health beliefs, culture and ethnicity in presentations of physical and psychological conditions
- Demonstrate how Individual behaviours impact on others; personality types, group dynamics, learning styles, leadership styles

Skills

- Develop a relationship that facilitates solutions to patient's problems
- Deal appropriately with behaviour falling outside the boundary of the agreed doctor patient relationship in patients, e.g. aggression, violence, sexual harassment
- Develop a self-management plan with the patient
- Support patients, parents and carers where relevant to comply with management plans
- Encourage patients to voice their preferences and personal choices about their care
- Use assessment, appraisal, complaints and other feedback to discuss and develop an understanding of own development needs.

Behaviours

- Recognise the duty of the medical professional to act as patient advocate
- Demonstrate:
    - Acceptance of professional regulation
    - Promotion of professional attitudes and values
    - Probity and the willingness to be truthful and to admit errors

<u>Recognising Own Limitations</u>

Knowledge

- Know the extent of one's own limitations and the limitations of self-professional competence and know when to ask for advice.
- Recognise that personal beliefs and biases exist and understand their impact (positive and negative) on the delivery of health services

Skills

- Reflection on individual practice.

Behaviours

- Be willing to consult and to admit mistakes.
- Be confident and positive in one's own professional values
- Be aware of one's own behaviour and how it might impact on patients' health issues

<u>Stress & Personal Health</u>

Knowledge

- Know the effects of stress and tools and techniques for managing it
- Demonstrate knowledge of the role and responsibility of occupational health and other support networks for doctors
- Know about one's responsibilities to the public

Skills

- Develop appropriate coping mechanisms for stress and ability to seek help if appropriate
- Demonstrate the ability to recognise the manifestations of stress on self and others and know where and when to look for support
- Balance personal and professional roles and responsibilities. Prioritise tasks, having realistic expectations of what can be completed by self and others

Behaviours

- Being conscientious, able to manage time and delegate
- Recognise personal health as an important issue
- Recognise personal health as an important issue.

Life-Long Learning

To inculcate the habit of life-long learning

Knowledge

- Define continuing professional development

Skills

- Recognise and use learning opportunities.
- Use the potential of study leave to keep oneself up to date

Behaviours

- Be:
    - self-motivated
    - eager to learn
- Show:
    - willingness to learn from colleagues
    - willingness to accept criticism

N. ETHICS AND LEGAL ISSUES

- Principles of Medical Ethics and Confidentiality
- To know, understand and apply appropriately the principles, guidance and laws regarding medical ethics and confidentiality

Knowledge

- Demonstrate knowledge of the principles of medical ethics
- Outline and follow the guidance given by the GMC on confidentiality

- Define the provisions of the Data Protection Act and Freedom of
- Information Act
- Define the principles of Information Governance
- Outline situations where patient consent, while desirable, is not required for disclosure e.g. serious communicable diseases, public interest
- Outline the procedures for seeking a patient's consent for disclosure of identifiable information
- Recall the obligations for confidentiality following a patient's death
- Recognise the problems posed by disclosure in the public interest, without patient's consent

- Recognise the factors influencing ethical decision making: including religion, personal and moral beliefs, cultural practices
- Outline the principles of the Mental Capacity Act
- Demonstrate an understanding of adolescents' and young adults'
- right to confidentiality and the importance of safeguarding

Skills

- Use and share information with the highest regard for confidentiality and encourage such behaviour in other members of the team, whilst recognising that the familial nature of genetics means that respecting individual confidentiality can be more complex.
- Use and promote strategies to ensure confidentiality is maintained e.g. anonymisation
- Counsel patients on the need for information distribution within members of the immediate healthcare team

Behaviours

- Encourage informed ethical reflection in others
- Show willingness to seek advice of peers, legal bodies, and the SLMC in the event of ethical dilemmas over disclosure and confidentiality
- Respect patient's requests for information not to be shared, unless this puts the patient, or others, at risk of harm
- Show willingness to share information about their care with patients,
- unless they have expressed a wish not to receive such information

<u>Informed Consent</u>

To ensure the trainee has the knowledge and skills to deal appropriately with ethical and legal issues that arise during the management of patients with genetic disorders.

Knowledge

- Know the process for gaining informed consent
- Understand process of consent for tissue/sample storage and use
- How to gain consent for a research project
- Outline the guidance given by the GMC on consent

Skills

- Present all information to patients (and carers) in a format they understand, checking understanding and allowing time for reflection on the decision to give consent with appropriate use of written material
- Provide a balanced view of all care options

Behaviours

- Respect a patient's rights of autonomy even in situations where their
- decision might put them at risk of harm
- Do not withhold information relevant to proposed care or treatment in a competent patient
- Does not seek to obtain consent for procedures in which they are not competent to perform, in accordance with GMC/regulatory Show willingness to seek advance directives

- Show willingness to obtain a second opinion, senior opinion, and legal advice in difficult situations of consent or capacity
- Inform a patient and seek alternative care where personal, moral or religious belief prevents a usual professional action

Legal Framework for Practice

To understand the legal framework within which healthcare is provided in the UK and/or devolved administrations in order to ensure that personal clinical practice is always provided in line with this legal framework

Knowledge
- All decisions and actions must be in the best interests of the patient
- Understand sources of medical legal information
- Understand disciplinary processes in relation to medical malpractice

Skills
- Ability to cooperate with other agencies with regard to legal requiremets
- Practice and promote accurate documentation within clinical practice

Behaviours
- Show willingness to seek advice from the employer, appropriate legal bodies (including defence societies), and the GMC on medico-legal matters
- Promote informed reflection on legal issues by members of the team
- All decisions and actions must be in the best interests of the patient

O. ORGANISATION AND PROVISION OF GENETICS SERVICES FOR POPULATIONS

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 of populations, common gene frequencies and disease prevalence
- The factors that influence decisions to instigate programmes of population screening for genetic diseases
- Define sensitivity, specificity, and predictive values of screening tests.
- Knowledge of current screening programmes
- Knowledge of appropriate population-based registers

Skills
- 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

Behaviours

- 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)

P. PATIENT EDUCATION AND DISEASE PREVENTION

Educating Patients about Disease, Investigations and Management
To ensure that the trainee has the knowledge, skills and attitudes to be able to educate patients effectively about genetic disease.

Knowledge

- Understand the genetic factors which influence the incidence and prevalence of common conditions
- Understand the factors which influence health and illness – psychological, biological, social, cultural and economic especially poverty
- Understand the influence of lifestyle on health and the factors that influence an individual to change their lifestyle
- Understand the purpose of screening programmes and know in outline the common programmes available within Sri Lanka
- Understand the positive and negative effects of screening on the individual
- Demonstrate in practice an appropriate knowledge of the influences of environment and behaviour on health including major factors such as poverty and poor housing, as well as those that might be overlooked

Skills

- Identify opportunities to promote changes in lifestyle and other actions which will positively improve health and/or disease outcomes.
- Identify the interaction between mental, physical and social wellbeing in relation to health
- Counsel patients appropriately on the benefits and risks of screening and health promotion activities
- Identify patient's ideas, concerns and health beliefs regarding screening and health promotions programmes and be capable of appropriately responding to these

Behaviours

- Encourage patients to access further information and patient support groups
- Engage in effective team-working around the improvement of health
- Encourage where appropriate screening to facilitate early intervention

<u>Managing Long-Term Conditions and Promoting Patient Self-Care</u>

Work with patients and use their expertise to manage their condition collaboratively and in partnership, with mutual benefit

To pursue a holistic and long-term approach to the planning and implementation of patient care, in particular to identify and facilitate the patient's role in their own care

Knowledge

- Describe the natural history of diseases and illnesses that run a chronic course
- Define the role of rehabilitation services and the multi-disciplinary team to facilitate long-term care
- Outline the concept of quality of life and how this can be measured whilst understanding the limitations of such measures for individual patients
- Outline the concept of patient self-care and the role of the expert patient
- Know, understand and be able to compare and contrast the medical and social models of disability
- Understand the relationship between local health, educational and social service provision including the voluntary sector.

Skills

- Develop and agree a management plan with the patient (and carers), ensuring comprehension to maximise self-care within care pathways where relevant
- Develop and sustain supportive relationships with patients with whom care will be prolonged and potentially life long
- Promote and encourage involvement of patients in appropriate support networks, both to receive support and to give support to others
- Encourage and support patients in accessing appropriate information

Behaviours

- Put patients in touch with the relevant agency including the voluntary sector from where they can procure items and other help as appropriate
- Show willingness to maintain a close working relationship with other members of the multi-disciplinary team, primary and community care
- Recognise and respect the role of family, friends and carers in the management of the patient with a long-term condition and the effect of full time caring on carer well-being

Q. WORKING WITH COLLEAGUES

Interactions Between: Hospital & GP, Hospital & Other Agencies e.g. Social Services Medical and Surgical Specialties
To demonstrate good working relationships with Colleagues

Knowledge

- Know the roles and responsibilities of team members and know how a team works effectively
- Know the role of multidisciplinary management in genetic disorders.
- The principles of effective inter-professional collaboration to optimise patient, or population, care
- Demonstrate knowledge of facilitation and conflict resolution methods

Skills

- Show leadership, delegate and supervise safely
- Be able to communicate effectively
- Recognise when input from another specialty is required for individual patients
- Be able to work effectively with GPs, other medical and surgical specialists and other health care professionals
- Employ behavioural management skills with colleagues to prevent and resolve conflict and enhance collaboration
- Demonstrate the ability to facilitate, chair, and contribute to meetings
- Prepare for meetings - reading agendas, understanding minutes, action points and background research on agenda items
- Maintain and routinely practice critical self-awareness, including able to discuss strengths and weaknesses with supervisor, recognising external influences and changing behaviour accordingly
- Create open and non-discriminatory professional working relationships with colleague's awareness of the need to prevent bullying and harassment
- Develop effective working relationships with colleagues and other staff through good communication skills, building rapport and articulating own view
- Communicate effectively in the resolution of conflicts, providing feedback, and identifying and rectifying team dysfunction

Behaviours

- Foster a supportive and respectful environment where there is open and transparent communication between all team members
- Ensure appropriate confidentiality is maintained during communication with any member of the team

R. TEACHING AND EDUCATIONAL SUPERVISION

To Have the Skills, Attitudes and Practices of a Competent Teacher
To demonstrate the knowledge, skills and attitudes to provide appropriate teaching, learning and assessment opportunities in Clinical Genetics for varied groups (medical, other health professional and lay groups).

Knowledge

- Outline the structure of an effective appraisal interview
- Differentiate between formative and summative assessment and define their role in medical education
- Outline the role of workplace-based assessments, the assessment tools in use, their relationship to course learning outcomes, the factors that influence their selection and the need for monitoring evaluation
- Outline the appropriate local course of action to assist a trainee experiencing difficulty in making progress within their training programme

Skills

- Be able to critically evaluate relevant educational literature
- Vary teaching format and stimulus, appropriate to situation and subject
- Provide effective feedback and appropriate after teaching, and promote learner reflection
- Conduct developmental conversations as appropriate e.g.: appraisal, supervision, mentoring
- Demonstrate effective lecture, presentation, small group and bed side teaching sessions
- Participate in strategies aimed at improving patient education e.g. talking and listening at support group meetings
- Be able to lead departmental teaching programmes including journal clubs

Behaviours

- In discharging educational duties acts to maintain the dignity and safety of patients at all times
- Recognise the importance of the role of the physician as an educator within the multi-professional healthcare team and uses medical education to enhance the care of patients
- Encourage discussions with colleagues in clinical settings to colleagues to share knowledge and understanding
- Maintain honesty and objectivity during appraisal and assessment
- Show willingness to participate in workplace-based assessments and demonstrates a clear understanding of their purpose
- Demonstrates a willingness to advance own educational capability through continuous learning
- Acts to enhance and improve educational provision through evaluation of own practice

S. RESEARCH

To be able to plan and analyse research

Knowledge

- Know how to use appropriate statistical methods
- Know the principles of gaining regulatory approvals for clinical research (Ethics, R and D approval, MHRA approval)
- Know how to analyse a scientific paper
- Outline the GMC guidance on good practice in research
- Understand the principles of research governance Outline the differences between audit and research
- Describe how clinical guidelines are produced
- Demonstrate a knowledge of research principles
- Outline the principles of formulating a research question and designing a project
- Comprehend principal qualitative, quantitative, bio-statistical and epidemiological research methods

Skills

- Undertake systematic critical review of scientific literature
- Ability to frame questions to be answered by a research project
- Develop protocols and methods for research
- Participate in collaborative research with clinical/scientific colleagues
- Be able to accurately analyse data
- Write and submit a case report or scientific paper
- Develop critical appraisal skills and apply these when reading literature

Behaviours

- Demonstrate curiosity and a critical spirit of enquiry
- Humility and the acknowledgement of the contribution of others
- Follow guidelines on ethical conduct in research and consent for research

T. CLINICAL GOVERNANCE

Demonstrate an understanding of the context, the meaning and the implementation of Clinical Governance.
The organisational framework for Clinical Governance at local, health authority and national levels.
Understanding of the benefits a patient might reasonably expect from Clinical Governance.
Creating an environment where mistakes and mismanagement of patients can be openly discussed and learned from.

Knowledge

- Know about quality improvement methodologies including a range of methods of obtaining feedback from patients, the public, and staff
- Know the principles and processes of evaluation, audit, research and development, clinical guidelines and standard setting in improving quality
- Outline a variety of methodologies for developing creative solutions to improving services

Skills

- Be an active partaker in clinical governance
- Assess and analyse situations, services and facilities in order to minimise risk to patients and the public

Behaviours

- Act as an advocate for the service
- Actively seek advice / assistance whenever concerned about patient safety
- Willing to take responsibility for clinical governance activities, risk management and audit in order to improve the quality of the service

<u>Evidence-Based Medicine</u>

Knowledge

- Know & understand the principles of evidence-based medicine
- Know & understand the types of evidence

- Understands of the application of statistics in scientific medical practice
- Understand the advantages and disadvantages of different study methodologies (randomised control trials, case-controlled cohort etc.)
- Understand the principles of critical appraisal
- Understand levels of evidence and quality of evidence
- Understand the role and limitations of evidence in the development of clinical guidelines and protocols
- Understand the advantages and disadvantages of guidelines and protocols
- Understand the processes that result in nationally applicable guidelines (e.g. NICE and SIGN)

Skills

- Able to critically appraise evidence
- Ability to be competent in the use of databases, libraries and the internet
- Able to discuss the relevance of evidence with individual patient.
- Ability to search the medical literature including use of PubMed, Medline, Cochrane reviews and the internet
- Appraise retrieved evidence to address a clinical question
- Apply conclusions from critical appraisal into clinical care
- Identify the limitations of research
- Contribute to the construction, review and updating of local (and national) guidelines of good practice using the principles of evidence-based medicine

Behaviours

- Display a keenness to use evidence in the support of patient care and own decisions therein.
- Keep up to date with national reviews and guidelines of practice (e.g. NICE and SIGN)
- Aim for best clinical practice (clinical effectiveness) at all times, responding to evidence-based medicine
- Recognise the occasional need to practise outside clinical guidelines

<u>Audit</u>

Knowledge

- Understand the different methods of obtaining data for audit including patient feedback questionnaires, hospital sources and national reference data
- Understand the role of audit (improving patient care and services, risk management etc.)
- Understand the steps involved in completing the audit cycle
- Understands the working and uses of national and local databases used for audit such as specialty data collection systems, cancer registries, etc. The working and uses of local and national systems available for reporting and learning from clinical incidents and near misses

Skills

- Involvement in on-going audit.
- Undertake at least one audit project.
- Design implement and complete audit cycles

- Contribute to local and national audit projects as appropriate
- Support audit by junior medical trainees and within the multidisciplinary team

Behaviours

- Recognise the need for audit in clinical practice to promote standard setting and quality assurance

Patient Safety

To understand that patient safety depends on the effective and efficient organisation of care, and health care staff working well together. To understand that patient safety depends on safe systems not just individual competency and safe practice. To understand the risks of treatments and to discuss these honestly and openly with patients so that patients are able to make decisions about risks and treatment options. Ensure that all staff are aware of risks and work together to minimise risk.
To recognise the desirability of monitoring performance, learning from mistakes and adopting no blame culture in order to ensure high standards of care and optimise patient safety

Knowledge

- Understand the elements of clinical governance
- Recognise that governance safeguards high standards of care and facilitates the development of improved clinical services
- Define local and national significant event reporting systems relevant to specialty
- Recognise importance of evidence-based practice in relation to clinical effectiveness
- Outline local health and safety protocols (fire, manual handling etc.)
- Understand risk associated with the trainee's specialty work including biohazards and mechanisms to reduce risk
- Keep abreast of national patient safety initiatives including National Patient Safety Agency, NCEPOD reports, NICE guidelines etc.
- Understands the investigation of significant events, serious untoward incidents and near misses
- Outline the components of effective collaboration and team working
- Describe the roles and responsibilities of members of the healthcare team

Skills

- Maintain a portfolio of information and evidence, drawn from your medical practice
- Reflect regularly on your standards of medical practice in accordance with SLMC guidance on licensing and re-registration
- Practise with attention to the important steps of providing good continuity of care
- Accurate attributable note-keeping including appropriate use of electronic clinical record systems

- Demonstrate leadership and management in the education and training of junior colleagues and other members of the healthcare team
- Lead and participate in interdisciplinary team meetings
- Provide appropriate supervision to less experienced colleagues

Behaviours

- Show willingness to participate in safety improvement strategies such as critical incident reporting
- Develop reflection in order to achieve insight into own professional practice
- Demonstrates personal commitment to improve their own performance in the light of feedback and assessment
- Engage with an open no blame culture
- Respond positively to outcomes of audit and quality improvement
- Co-operate with changes necessary to improve service quality and safety
- Encourage an open environment to foster and explore concerns and issues about the functioning and safety of team working
- Recognise limits of own professional competence and only practise within these.
- Recognise the importance of induction for new members of a team

U. STRUCTURE OF THE NATIONAL HEALTH SERVICES AND THE PRINCIPLES OF MANAGEMENT

Structure of the National Health Services (NHS) and the Principles of Management
To display knowledge of the structure and organisation of the NHS nationally and locally.

Knowledge

- Understand the local structure of NHS systems in your locality recognising the potential differences between in difference provinces
- Understand the structure and function of healthcare systems as they apply to your specialty
- Understand the consistent debates and changes that occur in the NHS including the political, social, technical, economic, organisational and professional aspects that can impact on provision of service
- Demonstrate knowledge of:
    - The structure, financing, and operation of the NHS and its constituent organisations
    - Ethical and equality aspects relating to management and leadership e.g. approaches to use of resources/ rationing; approaches to involving the public and patients in decision making
    - Business management principles: priority setting and basic understanding of how to produce a business plan
    - The requirements of running of a department, unit or practice relevant to the specialty
    - Efficient use of clinical resources in order to provide care
    - Commissioning, funding and contracting arrangements relevant to the specialty

- How financial pressures experienced by the specialty department and organisation are managed
- Relevant legislation (e.g. Equality and Diversity, Health and Safety, Employment Law) and local Human Resource policies
- The duties, rights and responsibilities of an employer, and of a co-worker (e.g. looking after occupational safety of fellow staff)
- Individual performance review purpose, techniques and processes, including difference between appraisal, assessment and revalidation
- The responsibilities of the various Executive Board members and Clinical Directors or leaders Demonstrate knowledge of organisational performance management techniques and processes

Skills

- Develop skills in managing change and managing people.
- Develop leadership skills to play a leading role in developing provincial genetic services.
- Develop interviewing techniques and those required for performance reviews.
- Participate in managerial meetings
- Take an active role in promoting the best use of healthcare resources
- Work with stakeholders to create and sustain a patient-centred service
- Employ new technologies appropriately, including information technology
- Demonstrate the ability to develop protocols &guidelines and implementation of these
- Analyse feedback and comments and, integrate them into plans for the service
- Use clinical audit with the purpose of highlighting resources required
- Identify trends, future options and strategy relevant to the specialty and delivering patient services
- Compare and benchmark healthcare services

Behaviours

- Recognise the importance of equitable allocation of healthcare resources and of commissioning
- Recognise the role of doctors as active participants in healthcare systems
- Respond appropriately to health service objectives and targets and take part in the development of services
- Recognise the role of patients and carers as active participants in healthcare systems and service planning
- Show willingness to improve managerial skills (e.g. management courses) and engage in management of the service
- Demonstrate:
    - Being prepared to accept responsibility
    - Showing commitment to continuing professional development which involves seeking training and self-development opportunities, learning from colleagues and accepting constructive criticism

- Commitment to the proper use of public money. Showing a commitment to taking action when resources are not used efficiently or effectively

V. INFORMATION TECHNOLOGY, COMPUTER ASSISTED LEARNING AND INFORMATION MANAGEMENT

To Demonstrate Good Use of Information Technology for Patient Care and
For Own Personal Development
Demonstrate competence in the use and management of health information.

Knowledge

- Know how to retrieve and utilize data recorded in clinical systems.
- Understanding the range of possible uses for clinical data and information and appreciate the dangers and benefits of aggregating clinical data.
- Skills
- Demonstrate competent use of database, word processing and statistics programmes
- Undertake effective literature searches
- Access genetic web sites and specialist databases to undertake searches
- Produce effective computer assisted presentations

Behaviours

- Be willing to offer advice to lay person on access to appropriate internet sources and support groups
- Adopt proactive and enquiring attitude to new technology
- Contribute to the development of sensitive validation frameworks to enable patients and their families to make judgements between different sources of information, advice and support

ANNEX 2 – FORMAT FOR THE REFLECTIVE LOG ACADEMIC/CLINICAL ACTIVITIES

Section 1:

1. Name of trainee: ……
2. Name(s) of supervisor(s): ……
3. Training centre/appointment: ……

Section 2:

4. Activity name ……
5. Date of activity: .....
6. List of expected learning outcomes:

   ……

   ……

   ……

   ……

7. Reflection (Please note that the questions indicated may not be applicable for all entries and therefore should be used only as a guide in relevant instances)

What … … is the situation/activity? … am I trying to achieve (your learning outcomes)? … actions did I take? … was the response of others? … were the consequences – for myself/for others?	
So, what … … does this teach me? … was I thinking and feeling? … other knowledge can I bring to the situation? … is my new understanding of the situation?	

Now what … … do I need to do to improve things? … broader issues need to be considered if this action is to be successful? … might I do differently in the future? … might be the consequences of this action?	

……
Signature of the trainee

……
Signature of the resource person/ clinical supervisor

Date: ……

Date: …….

Section 3:

Date of submission to PGIM : ……..

Date of approval by the Board of Study : ……..

Signature of Secretary of the Board of Study : ……..

Date ……..

ANNEX 3 – FORMAT FOR THE RESEARCH PROJECT PROPOSAL

Please make use of the following structure when preparing the research project proposal.

Section 1

1. Name of trainee
2. Name of supervisor
3. Training centre

Section 2

4. Project title
5. Background
6. Objectives of study
7. Methodology
    a. Design
    b. Subjects
    c. Laboratory Methods
    d. Bioinformatics
    e. Statistical Analysis
    f. Ethical considerations
8. Work plan and time lines
9. References
10. Funding for study
11. Signature of trainee

Section 3

Recommendation of supervisor
Signature of Supervisor
Date

Section 4

Date of submission to PGIM Date of approval by the Board of Study
Date
Signature of Secretary of the Board of Study

ANNEX 4 – CLINICAL TRAINING PROGRESS REPORT FORMAT

Section 1

1. Name of trainee : ……
2. Name of supervisor: ……
3. Training centre: ……..
4. Appointment: ……..

Section 2

5. Theoretical knowledge
   ❑ poor ❑ below average ❑ average ❑ Good ❑ Excellent

6. Clinical skills
   ❑ poor ❑ below average ❑ average ❑ Good ❑ Excellent

7. Clinical judgment
   ❑ poor ❑ below average ❑ average ❑ Good ❑ Excellent

8. Attitudes

   - Reliability
     ❑ poor ❑ below average ❑ average ❑ Good ❑ Excellent
   - Self motivation
     ❑ poor ❑ below average ❑ average ❑ Good ❑ Excellent
   - Team Leadership
     ❑ poor ❑ below average ❑ average ❑ Good ❑ Excellent
   - Teaching commitment
     ❑ poor ❑ below average ❑ average ❑ Good ❑ Excellent

9. Professional and ethical conduct
   ❑ poor ❑ below average ❑ average ❑ Good ❑ Excellent

Section 3

…….. ……..
Signature of the trainee Signature of the supervisor

Date : ……. Date: ……

Section 4

Date of submission to PGIM : ……

Date of approval by the Board of Study: ……

Signature of Secretary of the Board of Study: …….

Date: ……

ANNEX 5 – RESEARCH PROJECT PROGRESS REPORT FORMAT

Please note that section 1 of this report shall be completed by the trainee and the section 2 shall be completed by the supervisor.

Section 1: To be completed by the trainee

1. Name of Trainee: ……
2. Training Centre: ……
3. Supervisor: ……
4. Title of project: ……
5. Description of work carried out to date:

   Trainees self-assessment (Please describe progress in lab / field work and writing)

Section 2: To be completed by the supervisor

6. Is the work on schedule? Yes ☐ No ☐

7. Description of work carried out to date:
   Supervisors comments (Please comment on progress in lab / field work and writing)

8. Constraints (if any)

9. Recommendation of the supervisor: Satisfactory ☐ Unsatisfactory ☐

Date: ……. ……..
Signature of the supervisor

10. Recommendation of the BOS:

Date: ……..... ……..
Signature of Secretary:

ANNEX 6 - FORMAT FOR FORMATIVE ASSESSMENT

Name of the trainee	
Name of the trainer	
Assessment date	
Training Unit	

This section should be completed jointly by the trainer and trainee.

Area of competency	Marking Scale and Descriptors						Mark
	Poor	Average			Excellent		
	1	2	3	4	5	6	
A) Clinical Skills							
History taking	• Incomplete • Inaccurately • recorded	• Usually complete • Orderly and systematic			• Comprehensive and perceptive • Precisely recorded		
Physical examination	• Incomplete, inaccurate, lacks basic skills • Relies unnecessarily on investigations	• Thorough, confident examination • Recognizes most significant abnormalities			• Through and accurate • Knows and elicits specialist signs		
Investigations	• Inappropriate, random, unnecessarily expensive	• Usually appropriate			• Consistently appropriate in relation to differential diagnosis		
Judgement	• Unreliable • Does not grasp significance of clinical data • Fails to take appropriate action	• Reliable • Generally, interpret clinical data correctly. • Asks for advise appropriately			• Outstanding clinician, yet aware of his or her limits • Consistently correct decisions in complex cases		
Technical skills	• Slow to learn a technique • Unsatisfactory at routine procedures	• Reasonably quick to learn a technique • Routine procedures			• Learns rapidly • Routine procedures carried out fluently and manages		

	• Lacking confidence	carried out satisfactorily • Reasonably confident	difficult ones well • Very confident in technical skills	
Patient management	• Unsatisfactory at organizing data Misses important aspects of clinical problems • Formulates ineffective action plans	• Usually organizes data well • Considers most aspects of a problem • Action plans are usually effective	• Organises data very well • Has a clear view of problems • Action plans always compatible with the problem	
B) Knowledge				
Basic Science	• Uninterested, does not read the literature • Fails to apply basic science knowledge to clinical problems	• Reasonably up to date with the literature • Satisfactorily relates reading to patient care	• Avid reader of literature • Outstanding Knowledge of basic science applied well to patient care	
Clinical	• Not well read • Lack appropriate knowledge to construct a differential diagnosis • Does not learn from experience	• Reasonably well read • Satisfactory knowledge for dealing with common disorders, may miss some aspects of complex cases • Usually modifies practice in the light of experience	• Widely read, • Outstanding knowledge • Alert to unusual cases, seeks advice from senior colleagues to confirm observations • Constantly modifies practice according to experience	
C) Attitudes				
Reliability	• Unreliable • Forgets to carry out instructions	• Dependable • Conscientious in-patient care	• Thoroughly dependable, takes initiative • Anticipates problems and is willing to	

			discuss these with seniors	
Self-motivation	• Lacks enthusiasm and initiative • Minimal contribution to the team	• Contribution sound especially when encouraged by others • Actively involved in a team, contributes ideas	• Enthusiasm and initiative sustained even under duress • Good for moral when working with others	
Leadership	• Very limited, often alienates others • Colleagues and other staff confused by his or her instructions	• Has reasonable ability to influence others • Usually gives clear instructions	• Exceptional in directing and influencing others • Sets out clear guidelines and encourages others to take initiative	
Administration	• Always behind, badly organized	• Conscientious, quite well organised	• Excellent organiser, always on top of the work	
Colleagues	• Fails to get on with seniors, peers or juniors • Creates problems rather than solves them corporate with the workload of others	• Does not • Good rapport with seniors, peers and juniors • Sometimes a useful intermediary • Usually willing to help out others in a crisis	• Well respected by seniors, peers and juniors • Able to diffuse dissent amongst colleagues • Selfless, always to help even if personally inconvenient	
Other staff	• Disregards their skills: rude and unprofessional • Generates staff problems	• Respectful and appreciative of other staff, professional approach • Mediates when problems arise between professional groups	• Good rapport with other professionals, inspires enthusiasm • Professional and diplomatic if problems arise between groups	

Patients	• Poor at listening and communicating • Patients prefer other doctors • Increases anxieties	• Generally good at listening and communicating • Patients willing to be seen by him/her • Caring approach can allay fears	• Excellent at listening and communicating • Patients choose to be seen by him/her Inspires confidence	
D) Postgraduate Activities				
Teaching	• Uninterested and avoids teaching	• Competent and conscientious	• Excellent clinical teacher can inspire	
Lecturing	• Avoids if possible • Poorly prepared • Poorly delivered	• Regular participant • Good preparation • Good delivery	• Keen to lecture • Excellent preparation • Superb communicator	
Presentations	• Is not committed to giving papers • Poor presenter fails to extract and sequence the key features • Poor illustrations • Cannot respond appropriately to questions	• Enthusiastic presenter • Good delivery, well sequenced information • Appropriate graphic enhance delivery • Responds well to questions	• Inspirational presenter • Information sound with good interpretation • Outstanding illustrations • Responds to questions in a way that encourages development and original ideas	
Written communications	• Written style unclear and difficult to understand	• Written style usually clear giving unambiguous direction to others • Clear	• Written style appropriately adapted in vocabulary for the recipient	
Research ability	• Lacking in inclination to carry out research, not alert to opportunities	• Interested in research activities and has a reasonable	• Flair for original research • Well able to carry out research	

	• Unable to carry out directed projects	grasp of research methods • Requires supervision but competent when given direct support	independently and synthesises results well	
Audit	• Avoids if possible • Contributes little	• Regular attender Presents topics regularly	• Keen participant • Completes outstanding topics and implements the outcomes	

E) Comments by the trainer

(Summary of trainee's character and special attributes or failures. Mention any specific weakness that might hinder further training or requires special attention)

F) Comments by the trainee

(Mention any positive aspects or problems encountered during your current attachment. Include any unforeseen problems such as illness that might have affected your performance.)

……..	……..
Signature of the trainer	Signature of the trainee
Date: …….	Date: …….

ANNEX 7 – PRE-BOARD CERTIFICATION ASSESSMENT (PBCA) MARKING SCHEME

The overall assessment will be based on each of the main sections, which shall be assessed as satisfactory or not on an overall basis.

Domain	Expected Entries	Present/ Absent	Overall assessment
Subject expertise	Reflective logs related to academic activities taken part during training (as indicated in 6.1)		
	Reflective logs related to clinical training activities taken part during training (as indicated in 6.2)		
	Reflective logs pertaining to external events relevant to Clinical Genetics taken part during the training (as indicated in 6.1)		
	Five case reports in manuscript format or if a case has been published, the published manuscript where trainee is the first author.		
	Clinical training progress reports		
	Evidence of reading literature around Clinical Genetics including a brief reflection of learning		
	Formative assessments (conducted every six months)		
	Knowledge-based assessment reports (2 in number)		
Teaching	Evidence of undertaking teaching activities in areas related to Clinical Genetics for medical students, medical doctors or other health staff		
Research and audit	Published manuscript of the research project [In the case of an accepted manuscript awaiting publication: the manuscript, with the letter/email of acceptance]; or a detailed project report.		
	The Audit Report with evidence of presentation to a clinical meeting in the Institute (eg. Letter from the Head of the Institute) or a professional association (eg. Published abstract or if the abstract was not published, a letter from the association confirming presentation)		

	[Note: If the audit has been published, the published manuscript. The trainee should be the first author of any published manuscript.].		
Ethics and professionali s-m	Evidence of taking part in the 'Professionalism Strand of the PGIM' including a brief reflection on professionalism in medicine.		
	Multi-Source Feedback reports		
Information Technology	Participation in training programmes / workshops related to information technology.		
	Evidence of searching for information and application of findings in practice		
	Evidence for developing online content, presentations and other electronic content related to Clinical Genetics.		
Lifelong Learning	Participation in conferences and meetings		
	Membership and contribution to knowledge through academic/ professional organizations in Clinical Genetics in advancing research and practice.		
	Evidence of continued interest in a specific area in Clinical Genetics		
Reflective practice	Evidence of reflective learning		

Overall assessment and recommendation (Please comment)
