MASTER OF PATHOLOGY (MEDICAL GENETICS)

Human Genome Centre
School of Medical Sciences
Universiti Sains Malaysia
Health Campus
16150 Kubang Kerian
Kelantan

TRAINING CURICULUM FOR TRAINEES AND SUPERVISORS UNIVERSITI SAINS MALAYSIA

-2022-

MASTER OF PATHOLOGY (MEDICAL GENETICS)

1. **Aim**

A four year post-graduate training program in Medical Genetics.

2. **Background**

The School of Medical Sciences, USM, was established in 1979 to train undergraduate medical doctors. This evolved into developing post-graduate training in Medicine, which begun in 1987. Presently there are more than fifteen specialty post graduate training programmes being conducted by the School of Medical Sciences, USM.

Master of Pathology (Medical Genetics) was started in 2010 in Universiti Sains Malaysia (USM), Health Campus, Kubang Kerian, Kelantan with its first student graduated in Medical Genetics Pathology in 2014. This programme has been established and approved by Ministry of Higher Education (MOHE) and Ministry of Health (MOH), Malaysia to be incorporated into the Master of Pathology (MPath) Programme.

The aim of this programme is to produce genetic pathologists who can contribute to the multidisciplinary range of skills required within pathology services to aid in the diagnosis, management and treatment of patients with disorders arising from genomic mutations.

Currently, USM is the only centre in Malaysia offering this programme. Up till 2021, ten Genetic Pathology students have graduated and are now working in the hospitals under the MOH and public universities in Malaysia, whereas a total of fourteen candidates have enrolled in this programme.

3. **Structure of course** (Appendix I) The training is divided into 2 phases.

Phase I (Year 1): General Pathology in various sub- disciplines of pathology including TWO (2) weeks in Medical Genetics

At the end of the Stage 1 course the candidate will sit for an examination in General Pathology (Part 1 examination) and must pass this examination in order to proceed to the Stage 2 course.

Phase II (Year 2, 3 &4): MPath Medical Genetics trainee (based in Human Genome Centre)

3.1 Curriculum structure Phase I

The curriculum is divided into 2 parts.

a. General pathology in various sub-disciplines of pathology

b. Basic principles in medical genetics

Phase	Year	Curriculum and Training Place	Assessment
I	1	• General pathology in various sub- disciplines of pathology • Basic principles in medical genetics • Basic skills at medical genetics service • Log book	• Continuous supervisor assessment • End of posting examination at the end of posting at each disciplines • Professional examination I

3.2 Curriculum structure Phase II (specialty trainee in medical genetics)

Phase	Year	Curriculum and Training Place	Assessment
II	2, 3 &4	• Clinical responsibilities in medical genetics services • Case reports • Log book • Dissertation (starting in year 2) • Learning of medical genetics topic in packages. • Research activities, CPC and attending conferences • Posting to scheduled laboratories by rotation • Hematology Department, USM • Pathology Department, USM • To attend ward round and specialist clinics such as: • Hemato-oncology Unit • Pediatrics Clinic/ward • Posting at an external training center • Institute for Medical Research • Genetics Laboratory, Hospital Tunku Azizah, Kuala Lumpur • UKM Medical Molecular Biology Institute (UMBI), UKM • Paediatrics Department, Faculty of Medicine, UM	• Continuous supervisor assessment • Professional examination II

4. Mode of Teaching and Learning

Various modes will be used to achieve effective learning objectives.

- Notes, articles, references and audiovisual material will be prepared together with candidates.
- Lectures and seminar will be given in packages to cover the whole syllabus of medical genetics.
- Candidates are expected to learn primarily through the hands-on and job training in an independent and executing laboratory procedures and the duties in medical genetics.
- Preparation of case reports
- Actively participating in academic and research activities.
- Writing up of dissertation. The title must be specific. The length is about 15,000 words.

5. Supervision and progress reports

The medical school will appoint a qualified genetic pathologists and university lecturers experts in genetics to be a supervisor for each candidate. The supervisor is responsible for the progress report of the candidate.

6. Examination and Assessment

Assessment during the course is divided into 2 parts

- Continuous assessment
- Examination (annual and end-phase examinations)

6.1 Phase I assessment

6.1.1 Continuous assessment

a. coursework
b. supervisor report
c. case reports

6.1.2 Professional examination I……100%

MCQ & Essay……. 50%
OSPE……... 50%

6.2 Phase II (year 2, 3 & 4)

6.2.1 Continuous Assessment
a. log book
b. supervisor report

Candidates are required to prepare and conduct a project, submit a dissertation/manuscript and to submit a logbook of their course duties. The logbook, dissertation submission and completion of case presentations prior to Part 2 examination (1 Cytogenetics case presentation, 1 Molecular Genetics case presentation) as pre-requisite to sit for Part 2 examination

6.2.2 Professional examination (end of year 4) Theory:

Essay (Paper I and II)……45%

Practical: ……..45%

OSCE/OSPE 60%
Long practical 40%

Viva-voce:……10%

6.2.3 Repeat examination

a. Repeat examination after six months

A candidate may be allowed to repeat the examination after six months if he or she has an overall score of 50% or more but has failed either the theory OR the practical component

In this repeat examination, the candidate will be examined in the failed component and be given a viva-voce. The student must achieve satisfactory continuous assessment to be eligible to sit for examination.

The candidate is only allowed to repeat examination twice consecutively for the same component (theory or practical). Upon failure of the second repeat attempt, the candidate is required to repeat both theory and practical components after a period of 6 months to 1 year based on conjoint exam board decision.

b. Repeat examination after one year.

A candidate may be allowed to repeat the examination after one year if he has obtained an overall score of less than 50% OR has failed BOTH the theory and practical components of the Part 2 examination.

7. Entrance Criteria

7.1 Have a valid medical Degree from a university recognized by Malaysian Medical Council (MMC).
7.2 be registered with the MMC.
7.3 complete at least 3 years of medical service.
7.4 pass the entrance examination and/or
7.5 pass an interview.

All candidates must pass the entrance examination before he or she can be eligible for the interview for selection into the programme.

For foreign candidates, requirements a-e above are applied, plus

a. Possess a Temporary Practicing Certificate issued by the MMC before starting practice.
b. Undergo clinical or laboratory attachment at a minimum of 3 months before joining the programme with satisfactory supervisor report.
c. Proof of proficiency in the English language. Candidates must obtain a minimum score of 6.0 in IELTS or 550 in TOEFL (obtained within 2 years prior to date of enrolment)

The Entrance Examination:

a. The examination consists of TWO True-False Multiple Choice Question papers. Each paper consists of 60 questions with 5 responses.The first paper consists of basic anatomic pathology, forensic pathology, microbiology and immunology questions. The second paper consists of basic haematology, chemical pathology and genetic questions.
b. Marking system: A computerised marking system will be used. There will be minus marking of 0.5 marks for wrong answer. The minimum mark for each question is 0 (no carryover of negative marks).
c. The pass mark is 50% and the candidate will be called for an interview. Those who obtain a mark between 45 -49% may be considered for interview.
d. The result of the entrance exam is validfor 2 years.
e. The examination will be conducted annually by one of the conjoint universities.

8. Duration of Training

The maximum duration permitted to complete the course is seven years. The maximum duration permitted to complete the Stage 1 course is two years and the maximum duration permitted to complete the Stage 2 course is five years.

9. Curriculum and syllabus

Syllabus that will be used is attached (Appendix II & III). However, the syllabus will be updated from time to time in view of the progress in this field of specialty.

10. Academic and Teaching Staffs

a. All academic staffs at the School of Medical Sciences (from relevant department) will be involved in teaching activities. This is particularly in the phase I where the major input of basic sciences and general pathology.

b. Phase II and III will be particularly involved genetic pathologists and university lecturers experts in genetics.

c. Lecturer and student ratio of Lecturer: Student (1:2)

d. Visiting professor and/or visiting lecturer will be appointed in running the teaching packages in form of seminars.

11. Administrative committee

The Human Genome Centre, School of Medical Sciences USM will be responsible in organizing and monitoring the program, preparing teaching schedule and organizing seminars pertaining to the program.

12. Administration of Examination

The Medical School will coordinate and execute all examination. The result will be discussed at the Examination Board before approval by the Medical School Board and the Post-graduate University Board

Appendix I

Course Structure

<table>
<tr><th>STAGE 1</th><th colspan="3">STAGE 2</th></tr>
<tr><td><u>YEAR 1</u>
2 semesters
(48 weeks of T&L,log book)</td><td><u>YEAR 2</u>
2 semesters
(48 weeksT&L,in-service
training, log book, and research activity)</td><td><u>YEAR 3</u>
2 semesters
(48 weeks T&L, in-service
training, log book, and research activity)</td><td><u>YEAR 4</u>
2 semesters
(48 weeks T&L, in-service
training, log book, and research activity)</td></tr>
<tr><td></td><td>Semester 1</td><td>Semester 3</td><td>Semester 5</td></tr>
<tr><td rowspan="4">TCL: Lectures

SCL: Practical, Seminar, Case Study, Journal Critique.

SDL: Writing case book.

Rotation:
-10 weeks in every discipline of Pathology.
- Orientation week (1 week)
- Intensive course (2 weeks)
- Study leave (3 weeks)

Total= 46 weeks</td><td>SCL: in-service training in laboratory/ward/clinic/mortuary at University Hospital or MOH Hospital.</td><td>SCL: in-service training in laboratory/ward/clinic/mortuary at University Hospital or MOH Hospital.</td><td>SCL: in-service training in laboratory/ward/clinic/mortuary at University Hospital or MOH Hospital.</td></tr>
<tr><td>TCL: Research methodology and preparation of research proposal.

SCL: Practical, Seminar, Case Study, Journal Critique</td><td>SCL: Practical, Seminar, Case Study, Journal Critique</td><td rowspan="2">SDL : Carrying out research activity / writing case book

** Submission and assessment of dissertation, log book and case book to examiner(s).</td></tr>
<tr><td>SDL: Case book writing / research proposal writing</td><td>SDL : Carrying out research activity / writing case book
.</td></tr>
<tr><td>Semester 2</td><td>Semester 4</td><td>Semester 6</td></tr>
<tr><td>SCL: in-service training in laboratory/ward/clinic/mortuary at University Hospital or MOH Hospital.</td><td>SCL: in-service training in laboratory/ward/clinic/mortuary at University Hospital or MOH Hospital.</td><td>SCL: in-service training in laboratory/ward/clinic/mortuary at University Hospital or MOH Hospital.</td></tr>
<tr><td>PART 1 PROFESSIONAL EXAMINATION
(2 weeks)</td><td>SDL : Carrying out research activity / writing case book</td><td>SDL : Carrying out research activity / writing case book</td><td>ASSESSMENT of dissertation, log book and case book
and
PART 2 PROFESSIONAL EXAMINATION
(2 weeks)</td></tr>
</table>

*T& L = Teaching and Learning; TCL = Teacher-centered Learning; SCL = Student-centered Learning; SDL = Self-directed Learning; MOH = Ministry of Health

Appendix II

SYLLABUS FOR PHASE I (YEAR 1)

LEARNING OBJECTIVE

a. **General objective:**

The primary objective of the Stage I course is to attain basic knowledge in medical genetics.

b. **Specific objectives**:

i. Acquire basic theoretical knowledge in medical genetics.

ii. Acquire basic understanding of the common laboratory techniques involved in cytogenetic and molecular genetics tests.

iii. Acquire basic knowledge of common genetic disorders and competence in pedigree drawing and analysis.

COURSE CONTENT

a. BASIC KNOWLEDGE OF MEDICAL GENETICS

List of lectures:

Title	**Hour**
Introduction to medical genetics	1
Introduction to cytogenetics	1
Protein synthesis	1
Genetic disorders and modes of inheritance	1
Genetic basis of human cancer	1
Application of cell culture in genetic research	1
PCR technology and its application in medical genetics	1
Mitochondrial disorders	1
Basics of bioinformatics	1
Cytogenetic analysis- methods and application	1
Pedigree analysis and drawing	1
Dysmorphic disorders	1
Genetic testing in cancer	1
Targeted therapy: predictive and prognostic biomarkers	1

Cancer cytogenetics	1
Genetic counselling and its scenario in Malaysia	1
Introduction to Variant Effect Prediction (VEP)	1
Pharmacogenomics	1

*topic of lectures might be varied or updated according to current needs

b. LABORATORY TECHNICAL COMPETENCIES

Practical Session:

Skills	**Hour**	**Competence Level**
Pedigree drawing and analysis	2	1
Fluorescence *in situ* hybridization (FISH) analysis	2	1
Chromosomal analysis (Karyotyping)	2	1
DNA extraction & quantification	2	1
Polymerase chain reaction (PCR)	2	1
Gel electrophoresis	2	1

TEACHING PROGRAMME

a. There will be formal sessions in the form of teaching and small group discussions & student presentation on selected topics.
b. Practical sessions will be held in genetic laboratory for conventional cytogenetic, molecular cytogenetic and molecular genetic techniques.

LEARNING OUTCOMES

a. Candidates should be able to understand the basic concepts in medical genetics and its clinical diagnostic applications.
b. Candidates are expected to understand the basic mechanism in genetic disorders.
c. Candidates should understand the basic principles and applications of common cytogenetic and molecular tests.

RECOMMENDED TEXTBOOKS/JOURNALS/REFERENCES

a. Emery's Elements of Medical Genetics and Genomics (16th edition, 2020) by Peter D Turnpenny, Sian Ellard and Ruth Cleaver (editors), Elsevier Churchill Livingstone.
b. Human Molecular Genetics (5th edition, 2019) by T. Strachan and Andrew Read. ISBN: 10.0815345895/ 13.9780815345893.
c. Thompson & Thompson Genetics in Medicine (8th edition, 2015) by Robert L Nussbaum, Roderick R. McInnes and Huntington P. Willard. ISBN: 978 - 1- 4160- 3080-5.
d. An International System for Human Cytogenomic Nomenclature (2020) by Jean McGowan-Jordan, Ros J. Hastings and Sarah Moore. ISBN: 978-3318067064.

Appendix III

SYLLABUS FOR PHASE II (Year 2, 3 and 4)

INTRODUCTION

The Stage 2 course of Master of Pathology (Medical Genetics) is THREE (3) years duration. The candidates will be placed at the Human Genome Centre, School of Medical Sciences, USM. There are three important aspects which are included during the programme; theoretical, practical and research.

Laboratory attachments will be carried out in Human Genome Centre, USM, Genetic Laboratory, Hospital Tunku Azizah, Kuala Lumpur as well as other institutions in Malaysia (please refer to the Course Structure).

LEARNING OBJECTIVE

a. **General objective**:
The primary objective of the Stage 2 programme is for the candidate to attain advanced knowledge and practical competence to direct and manage a genetic laboratory.

b. **Specific Objectives**:
To acquire consultant level skills in Medical Genetics in the following clinical and laboratory aspects:

i. Understand and attain competency with ability to provide consultancy services in cytogenetic and molecular genetic tests.
ii. Familiarize with various genetic disorders and conversant with the basic concept of genetic counseling.
iii. Gain expertise in test design, validation, and proper quality control with competency in laboratory management based on the concept of Laboratory Quality Management (LQM).
iv. Competent in the assessment, troubleshooting and interpretation of laboratory data and bioinformatics.
v. To be able to conduct a research project and familiarize with bioinformatics tools.
vi. Able to be innovative and adapt to the continuously evolving clinical needs in terms of diagnostic, therapeutics and personalized state of the art genomics in the Malaysian perspective.

COURSE CONTENT

The course covers the theoretical component, laboratory technical skills, laboratory management and research component.

a) THEORETICAL COMPONENT (Tutorials/ seminars/ lectures)

	TOPIC
Year 2	
1.	Introduction to medical genetics
2.	Introduction to molecular genetics
3.	Introduction to human cytogenetics
4.	Cytogenetic analysis – Methods and Applications
5.	Principles of inheritance in genetic disorders
6.	Introduction to genetic counselling
7.	Overview of DNA technology and applications in medicine
8.	Cancer genetics (molecular and cytogenetic techniques)
9.	Genetic Basis of human cancer
10.	Introduction to clinical genetics
11.	Introduction to cell culture
12.	Introduction to cytogenetics and molecular cytogenetics - Prenatal diagnosis - Postnatal diagnosis - Cancer cytogenetics - Other tissue cytogenetics
13.	Common molecular techniques in genetic tests
14.	Introduction International System for Human Cytogenomics Nomenclature
15.	Quality Management System Laboratory
16.	Ethics in human research
Year 3	
17.	Advanced Molecular Techniques for Diagnosis of Genetic Disorder - Microarray - NGS - Droplet digital PCR
18.	Cancer genetics - Solid tumours - Haematological cancer
19.	Population Genetics & Epidemiology
20.	Bioinformatics
21.	Congenital Heart Defect/ Cardiogenetics
22.	Mitochondrial Genetics & Neurogenetics
23.	Genetic counselling/Risk assessment
24.	Epigenetics
25.	Introduction to stem cell & its application
26.	Pharmacogenomics
27.	Birth Defects
28.	Introduction to gene therapy
29.	Personalized medicine and precision medicine
30.	Ethical and soft skills management
31.	Use and misuse of genetic tests

32.	Introduction to biobanking
33.	Non-invasive technique (liquid biopsy) in genetic testing

b) LABORATORY TECHNICAL SKILLS

Practical Skills to Be Acquired	Level of Competence
Analysis and hereditary pedigree	5
Karyotyping for whole blood and bone marrow samples	5
Fluorescence in-situ Hybridization (FISH) analysis	5
DNA extraction and quantification	5
RNA extraction and quantification	3
Polymerase chain reaction (PCR)	5
Genotyping/Mutational analysis using PCR-RFLP	5
Gel electrophoresis	5
DNA sequencing analysis	2
Mutational screening using dHPLC	2
Multiplex Ligation-dependent Probe Amplification (MLPA)	4
Real-Time quantitative PCR	4
Array Comparative Genomic Hybridization (aCGH)	2
Microarray and gene expression analysis	2
Blot assays (optional)	1

c) LABORATORY MANAGEMENT

i. Quality Management System (QMS) Laboratory

- MS ISO 15189 accreditation for Cytogenetics and Molecular Genetics Laboratory
- External Assurance Programme

d) RESEARCH COMPONENT

ii. Designing and conduct of research project
iii. Submission of dissertation / manuscript
iv. Presentation at scientific meeting or publication in indexed journals

COURSE STRUCTURE

Year 2

Scope/Content	Duration (Week)	Placement
Orientation	1	Human Genome Centre, USM
Introduction of Medical/ Human Genetics	4	
Research Methodology and Medical Statistics	3	School of Medical Sciences, USM
Introduction to Laboratory Quality Management System & Medical Genetic Testing	2	Human Genome Centre and/or Advanced Medical & Dental Institute, Bertam, USM
Postnatal Diagnosis Part 1 (blood & bone marrow) (Chromosomal & DNA analysis)	8	Human Genome Centre, USM
Clinical Genetics Part 1 Genetic Disorders in Pediatrics (Postnatal)	12	Human Genome Centre, USM
Clinical Genetics Part 2 Genetic Disorders in Pediatrics (Postnatal)	8	Paediatric Department, USM
Genetic Counselling Part 1	4	Paediatric Department/ Human Genome Centre, USM
Clinical Genetics Part 1 Genetic Disorders in Hematology	6	Hematology Department, USM
Holidays	4	

Year 3

Scope/Content	Duration (Week)	Placement
Cancer Genetics - Solid tumours	2	Pathology Department, USM
Cancer Genetics - Solid tumours - Hemato-oncology	8	Paediatrics Department/ Human Genome Centre USM
Cancer Genetics - Hemato-oncology	8	Internal Medicine/ Hematology Department, USM
Cancer Genetics - Hemato-oncology	2	Pathology Department, Faculty of Medicine, UKM
Advanced Molecular Techniques Part 1	2	UKM Medical Molecular Biology Institute (UMBI), UKM
Population Genetics	2	Human Genome Centre, USM
Genetic Epidemiology	2	Human Genome Centre, USM
Bioinformatics Application in Medical Genetics	4	Human Genome Centre, USM
Ethical Issues	2	Human Genome Centre, USM
Clinical Genetics Part 2 - Cardiogenetics - Neurogenetics - Reproductive genetics	6	Human Genome Centre/ School of Medical Sciences, USM
Clinical Genetics Part 3 - Neuropsychiatric genetics	2	Psychiatric Department, Hospital Kuala Lumpur
Genetic Counselling Part 2	4	Paediatrics Department, Faculty of Medicine, UM
Application of Laboratory Quality Management System & Medical Genetic Testing Part 1	4	Human Genome Centre, USM
Holidays	4	

Year 4

Scope/Content	Duration (Week)	Placement
Prenatal Diagnosis Part 1	4	Human Genome Centre, USM
Prenatal Diagnosis Part 2	2	Department of O&G, USM
Postnatal Diagnosis Part 2 - Chromosomal analysis - Molecular genetic analysis	30	Genetics Laboratory, Hospital Tunku Azizah, Kuala Lumpur Human Genome Centre, USM
Advanced Molecular Techniques Part 2	2	Institute for Medical Research (IMR, Kuala Lumpur) Human Genome Centre, USM Hospital Tunku Azizah, Kuala Lumpur
Mitochondrial Genetics	4	Human Genome Centre, USM Specialised Diagnostic Centre, Institute for Medical Research (IMR, Kuala Lumpur)
Application of Laboratory Quality Management System & Medical Genetic Testing Part 2	4	Human Genome Centre, USM
Revision	2	Human Genome Centre, USM
Professional Examination II	2	School of Medical Sciences, USM
Holidays	2	

TEACHING PROGRAMME

a. Lecturer and student ratio of Lecturer: Student (1:2)

b. Lectures will be held in the form of formal lectures, tutorials, laboratory practical, seminars, journal reviews and formal discussions.

c. The candidates are expected to learn primarily through the hands-on and on job training in an independent. All units/departments involved in this program has an approved accreditation by “Jawatankuasa Bersama Pengkhususan Patologi” committee

d. Candidates will be posted at all the scheduled lab posting by rotation

e. Candidates may be posted outside the mother university to other institutions
   e.g., Institute for Medical Research, Genetics Laboratory, Hospital Tunku Azizah, Kuala Lumpur or any other universities when deemed necessary in order to acquire necessary skills and exposure.

f. Candidates are encouraged to attend ward round and specialist clinics such as Hemato-oncology Unit, Pediatrics Clinic/ward, or other clinics which are deemed important.

g. Candidates are required to submit a logbook of their course duties. The logbook is a pre-requisite to sit for the Part 2 examination.

h. General teaching methodology:

   i. Several teaching methodologies will be carried out to ensure efficiency in teaching and learning.
   ii. A detailed synopsis of each lecture will be prepared. Appropriate list of books, magazines and monograph will be provided by the university.
   iii. The university will provide the appropriate lecture notes, articles and manuscripts from the medical journals and magazines, reference books and audio-visual materials as the teaching materials.
   iv. The schedule for lectures, tutorials, demonstrations, and laboratory practical will be allocated
   v. Candidates are required to attend selected posting in different laboratories and clinics/wards as listed in the course structure.
   vi. Candidates are required to prepare and conduct a project and submit a dissertation/manuscript as pre-requisite to sit for Part 2 examination.
   vii. Candidates are expected to participate actively in the Continuous Medical Education (CME) presentations, weekly Cytogenetics meeting, monthly Molecular Genetics meetings, journal club presentations, seminars, conferences and other teaching and learning activities.
   viii. The university will decide other appropriate teaching methodologies when deemed necessary.

Formative assessment

A. Logbook

i) Candidates will be required to maintain a logbook to ensure the expected training and self-learning process are achieved. The logbook must be reviewed by the supervisor.

ii) The completion of logbook is a pre-requisite to sit for the Part 2 examination

iii) The ownership of the logbook belongs to the university. Candidates will be posted routinely at other laboratories and centres outside the Human Genome Centre, USM, to acquire experience of different cases, facilities, and expertise.

iv) The following must be recorded in the logbook:

- Place of training
- A list of patients examined
- A list of interpretations of laboratory tests done
- Candidates are required to submit; as provided in the table format below:

Type	Quantity
Karyotype blood sample	150
Karyotyping on bone marrow	150
Molecular cytogenetics/FISH	100
External quality assurance programme	2

For Molecular Genetic testing, the testing involved are:

Test	Quantity
PCR-RFLP	20
PCR- DNA sequencing	20
Multiplex PCR	15
MLPA	10
Fragment Analysis	10
RT-PCR	10
Array CGH/DNA Microarray	10
Next-generation sequencing (NGS)	5

v) Presentation at scientific meetings must be recorded

vi) Publications (Submitted or published at least one local or international journal publications/case report prior to end of Year 4)

vii) Attendance at Cytogenetics meetings and Molecular Genetics meeting, and CME/Journal Club presentation

viii) Case presentation shall be completed prior to Part 2 examination (1 Cytogenetics case presentation, 1 Molecular Genetics case presentation)

LEARNING OUTCOMES

i. Candidate has sufficient knowledge in genetic disorders, genetic counseling, and the principles of various genetic tests.
j. Candidate is competent in the performance of various cytogenetic and molecular genetic testing.
k. Candidate becomes a specialist in test design, validation, and proper quality control.
l. Candidate is able to provide consultancy in laboratory diagnosis and expert interpretation of the test result.
m. Candidate is competent in laboratory management and quality control.
n. Candidate should be able to undertake research project independently.
o. Candidate is professional, ethical, holistic, adaptive, marketable and an effective communicator

RESEARCH PROJECT

Candidates are required to plan, undertake a research project, and write up a dissertation which has to be submitted 6 months prior to Part 2 examination. Dissertation will be examined by 2 internal examiners and reviewed by external examiner appointed by the university.

RECOMMENDED TEXTBOOKS/JOURNALS/REFERENCES

a) BOOKS

i. Emery's Elements of Medical Genetics and Genomics (16th edition, 2020) by Peter D Turnpenny, Sian Ellard and Ruth Cleaver (editors), Elsevier Churchill Livingstone.
ii. Human Molecular Genetics (5th edition, 2019) by T. Strachan and Andrew Read. ISBN: 10.0815345895/ 13.9780815345893.
iii. Thompson & Thompson Genetics in Medicine (8th edition, 2015) by Robert L Nussbaum, Roderick R. McInnes and Huntington P. Willard. ISBN: 978 - 1- 4160- 3080-5.
iv. An International System for Human Cytogenomic Nomenclature (2020) by Jean McGowan-Jordan, Ros J. Hastings and Sarah Moore. ISBN: 978-3318067064.
v. Arsham, M.S., Barch, M.J. and Lawce, H.J. eds., 2017. The AGT cytogenetics laboratory manual. John Wiley & Sons.
vi. Leonard, D.G., Bagg, A., Caliendo, A.M., Deerlin, V.M. and Kaul, K.L. eds., 2016. Molecular pathology in clinical practice. Cham, Switzerland: Springer International Publishing.
vii. Zneimer, S.M., 2014. Cytogenetic Abnormalities: Chromosomal, FISH, and Microarray-Based Clinical Reporting and Interpretation of Result. John Wiley & Sons.
viii. WHO classification of tumours of haematopoietic and lymphoid tissues / edited by Steven H. Swerdlow, Elias Campo,Nancy Lee Harris, Elaine S. Jaffe, Stefano A. Pileri, Harald Stein, Jurgen Thiele. - Revised 4th edition

b) **WEBSITES**

i. RCPA Education Online http://www.rcpa.edu.au/Education (specifically the Ethics,Quality Management and Laboratory Safety eLearning modules)
ii. RCPA website: http://www.rcpa.edu.au/Education/Disciplines/Genetic-Pathology
iii. European Molecular Genetics Quality Network website: http://www.emqn.org/emqn/Best+Practice (Accessed December 2015)
iv. Australasian Society of Diagnostic Genomics (ASDG) website: https://www.hgsa.org.au/asdg (Accessed October 2020).
v. The National Center for Biotechnology Information (NCBI) website: https://www.ncbi.nlm.nih.gov/ (Accessed October 2020).
vi. Emsebl website: https://ensemblgenomes.org/.

c) INFORMATION ABOUT RARE GENETIC TESTS

a. NIH Genetic Testing Registry – http://www.ncbi.nlm.nih.gov/gtr/ (Accessed December 2015)
b. NCBI Gene Reviews - http://www.ncbi.nlm.nih.gov/books/NBK1116/(Accessed December 2015)
c. EuroGentest – http://www.eurogentest.org/(Accessed December 2015)

d) QUALITY ASSURANCE/ BEST PRACTICE GUIDELINES

a. American College of Medical Genetics Standards and Guidelines for Clinical Genetics Laboratories (Accessed December 2015)
b. European Molecular Genetics Quality Network: http://www.emqn.org/emqn/Best+Practice (Accessed December 2015)
c. Association for Clinical Genetic Science (part of the federated British Society for Genetic Medicine):http://www.acgs.uk.com/committees/quality-committee/best-practice-guidelines/ (Accessed December 2015)
d. Swiss Society of Medical Genetics: http://sgmg.ch/wordpress/wp-content/uploads/2015/09/SGMG_Reporting_Guidelines.pdf(accessed December 2015)
e. Human Genetics Society of Australasia Policies, Guidelines and Position Statements (accessed December 2015)
f. NPAAC Guidelines: http://www.health.gov.au/internet/main/publishing.nsf/Content/health- npaac-publication.htm(accessed December 2015)
g. European Research Network for evaluation and improvement of screening, Diagnosis and treatment of Inherited Disorders of Metabolism (ERNDIM) (Accessed December 2015)