



COURSE SPECIFICATION

2nd part MD Pediatrics–Elective Genetics and Metabolism

Faculty of Medicine– Mansoura University

(A) Administrative information

(1) Programme offering the course:	MD Pediatrics (PED 600)
(2) Department offering the programme:	Pediatrics
(3) Department responsible for teaching the course:	Pediatrics
(4) Part of the programme:	2 nd part – Elective course Genetics and Metabolism
(5) Date of approval by the Department's council	27/4/2016
(6) Date of last approval of programme specification by Faculty council	9-8-2016
(7) Course title:	Elective course MD Genetics and Metabolism
(8) Course code:	PED 609 AGM
(9) Total teaching hours:	45 hours

(1) Course Aims:

The broad aims of the course are as follows. This course aims

To develop the knowledge, required to deliver clinical genetic services.

To develop skills in diagnosing genetic diseases using clinical evaluation and genetic testing, and choosing appropriate genetic investigations and interpreting results.

(2) Intended Learning Outcomes (ILOs):

On successful completion of the course, the candidate will be able to:

A. Knowledge and Understanding

- A1. Identify the principles of genetics and how to deal with common genetic disorders
- A2. Identify non-Mendelian modes of inheritance and principles of, mutations, microdeletion, Peroxisomal disorders, mitochondrial disorders, hypoglycemic syndromes and genetic causes of mental retardation.
- A3. Identify dysmorphic features in relation to common genetic syndromes and principles of genetic counseling.
- A4. Discuss methods of molecular and prenatal diagnosis of genetic disorders.
- A5. Identify and differentiate the features of metabolic disorders particularly organic acidemias, hyperammonemias, peroxisomal and mitochondrial disorders and hypoglycemic syndromes and how to manage.
- A6. Discuss recent trends of molecular diagnosis and flow cytometry.
- A7. Recognize characteristic features of common mutation disorders in pediatrics.

B. Intellectual skills

- **B1.** Design a practical approach to hyperammonimia
- **B2.** Design a family tree for genetic counseling
- **B3.** Compare methods of molecular diagnosis in different disorders
- **B4.** Develop an approach to microdeletion syndromes.
- **B5.** Interpret results of flow cytometry

(3) Course content:

Elective Pediatric MD Genetics and Metabolism

Course	Title	Teaching hrs	
		lectures	ILOs
Genetics and Metabolism	Non-Mendelian inheritance	3	A1
	Dysmorphic child	3	A3
	Mental Retardation	3	A2
	Genetic counselling	3	A4-B2
	Prenatal diagnosis	3	A4
	Mutations	3	A7
	Methods of molecular diagnosis 1	3	A6-B3
	Methods of molecular diagnosis 2	3	A6-B3
	Microdeletion syndromes	3	A2-B4
	Hyperammonemias	3	A2-B1
	Perioxosomal disorders	3	A2
	Mitochondrial disorders	3	A2
	Hypoglycemic syndromes	3	A5
	Advances courses in Molecular diagnosis	3	A6-B3
	Advances courses in flow cytometry	3	A6-B3-B5
Total		45	

(4) Teaching methods:

- 4.1. Weekly lectures and scientific seminar
- 4.2. Self learning

(5) Assessment methods:

- 5.1....Written for assessment of (knowledge, intellectual skills)
- 5.2.... Oral ... for assessment of.....(knowledge, intellectual skills)
- 5.3 MCQ for assessment of..... (knowledge, intellectual skills)

Assessment schedule : Semester & Final Exam.

Course	Marks		
	MCQ (Semester Exam)	Essay (Final Exam)	Oral
Genetics and Metabolism	12	48	30
Total	90		

(6) References of the course :

- Nelson's Text book of Pediatrics
- Emry textbook of genetics.
- Thompson and Thompson textbook of genetics.
- Smith's textbook of genetics

(7) Facilities and resources mandatory for course completion:

- **LECTURE HALLS:** One hall for lecturers is available at Mansoura University Children's Hospital (MUCH). The hall is equipped with white board, data show, and computer.
- **CLINICAL ROUNDS HALLS:**
One hall for clinical rounds is available at Mansoura University Children Hospital (MUCH). Computer and AV aids facilities are available with prior arrangement.
- **LIBRARY:**

The library is located on the 4th floor of the Faculty of Medicine, Mansoura University.

- **FACILITIES FOR TUTORS**

In addition to the library on 4th floor of the Faculty of Medicine, Mansoura University, there is a specialized pediatric library at MUCH (Professor Mohammad Hafez's Library).

The offices of all staff at MUCH are equipped with computers and high speed internet connection.

International databases are available through the website of the university (www.mans.edu.eg)

Course coordinator: Dr. Yahya Wahba

Head of the department: Prof. Ali Shaltout

Date: