



COURSE SPECIFICATION

Human Genetics

Faculty of Medicine– Mansoura University

(A) Administrative information

(1) Programme offering the course.	M.D. degree of Histology & Cytology
(2) Department offering the programme.	Histology & Cell biology
(3) Department responsible for teaching the course.	Histology & Cell biology
(4) Part of the programme.	First part
(5) Date of approval by the Department`s council.	30/4/2016
(6) Date of last approval of programme specification by Faculty council.	9-8-2016
(7) Course title.	Human Genetics
(8) Course code.	HIST 602 GE
(9) Credit hours	3
(10) Total teaching hours.	45

(B) Professional information

(1) Course Aim.

The aim of this course is to provide an in-depth background in all of the areas of genetics in order to enable the student to demonstrate the broad range of knowledge and proficiency that will help him to successfully move on and master advanced topics in cytogenetics and genetic engineering.

(2) Intended Learning Outcomes (ILOs).

A- Knowledge and Understanding

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

- A1 Describe the structure of DNA, RNA and chromosomes.
- A2 Explain the process of DNA replication and transcription.
- A3 Define the importance of telomere and centromere areas of chromosomes.
- A4 Recognize the mechanism of RNA processing and evaluate its importance.
- A5 Describe the genetic code and the steps of protein synthesis.
- A6 Identify gene expression and explain the mechanism of its regulation.
- A7 Explain DNA repair systems and identify the impact of a defective DNA repair.
- A8 List the components of the human genome and compare it with other genomes.
- A9 Identify mutation and explain its different causes, mechanisms and effect.
- A10 Identify the causes of genetic variation in natural population.
- A11 Explain the cell cycle and define the phases of interphase, mitosis and meiosis.
- A12 Classify genetic disorders, explain their causes and give examples for each type.
- A13 Discuss karyotyping and identify Barr's Body focusing on its significance
- A14 Explain the different patterns of inheritance, and the possible disorders.
- A15 Discuss the genetic basis of cancer.
- A16 Define restriction endonucleases and explain their role in molecular genetics.
- A17 Discuss the role of electrophoresis in molecular genetics.
- A18 Identify gene amplification and explain the mechanism of polymerase chain reaction.
- A19 Recognize and compare the different types of cloning vectors.
- A20 Explain the strategies of genetic engineering and some of its applications.
- A21 Identify blotting, hybridization and micro-array techniques.

- A22 Discuss DNA sequencing
- A23 Explain the technique and importance of finger and foot printing.
- A24 Discuss applications of gene therapy

B- Intellectual skills

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

- B1 Use karyotyping and Barr's bodies for the diagnosis of chromosomal anomalies.
- B2 Demonstrate chromosomes by different banding techniques,
- B3 Construct and analyze family pedigrees to detect the pattern of disease inheritance.
- B4 Differentiate DNA and RNA particles by centrifugation techniques.
- B5 Apply blotting techniques to separate DNA or RNA fragments.
- B6 Apply hybridization techniques to obtain a target DNA or RNA fragment.
- B7 Use cloning vectors or PCR to amplify genes.
- B8 Use the Sanger's technique to obtain the sequence of a gene or a DNA fragment.

(3) Course content.

Subjects	Lectures
<p>1- Basic Genetics</p> <ul style="list-style-type: none"> • Chemical & Physical Structure of DNA. • Structure of Chromosomes • Structure of Centromere & Telomere. • DNA Replication. • DNA Repair System. • DNA Transcription. • Structure & Types of RNA. • RNA Processing. • Regulation of Gene Expression. 	<p>13</p>

<ul style="list-style-type: none"> • The Genetic Code & Protein Synthesis. • Mutation. 	
2- Genomes <ul style="list-style-type: none"> • The Human Genome & Transposable Elements. • Genetic Variations in Natural Population. • The Genome of Microorganisms. • The Mitochondrial Genome. 	6
3- Cell Division, Karyotyping & Chromosomal Abnormalities <ul style="list-style-type: none"> • Interphase & Cell Division (Mitosis & Meiosis). • Karyotyping & Barr's Body. • Chromosome Banding Techniques. • Chromosomal Anomalies (Numerical & Structural). • The Mendelian Principles of Inheritance & Some Mendelian Disorders. • Family Pedigree & Chi-square Test. • The Genetics of Cancer. 	13
4- Genetic engineering <ul style="list-style-type: none"> • Restriction Endonucleases. • Gel-electrophoresis. • Blotting Techniques (Southern, Northern & Western). • Hybridization (ISH & FISH) & Micro-array Techniques. • Gene amplification (Cloning Vectors & PCR). • DNA Sequencing. • Gene Mapping. • Strategies of Genetic Engineering & Some Industrial Applications. • Finger & Foot-Printing. • Gene Therapy. 	13
Total teaching hours	45

(4) Teaching methods:

- 4.1: Lectures with data show and instructive videos.
- 4.2: Discussion and class activities.
- 4.3: Problem-based exercises.
- 4.4: Seminars.
- 4.5: Self learning (internet data collection).

(5) Assessment methods:

- 5.1: Assessment MCQ Exam: 20%: 20 Marks:
- 5.2: Final Written Exam: 80%: 80 Marks

(6) References of the course:

6.1: Course Notes: Basics of Genetics prepared by Professor Salwa Gawish.

6.2: Text books:

- Genetics From Genes to Genomes. Hartwell, Hood, Goldberg, Reynolds, Silver, Veres. 3rd Edition (2008); McGraw Hill.
- Principles of Genetics. Snustard & Simmons. 4th edition(2005); Wiley.
- Essentials of Genetics. Klug & Cummings. Fifth edition (2005); Pearson Education.

6.3: Journals:

- MUTATION RESEARCH
- CHROMOSOMA
- AM J HUM GENET
- CLIN GENET
- GENETICS
- J MED GENET

6.4: Websites:

- Jackson Laboratory
- Pubmed
- Genetic Science Learning Center

(7) Facilities and resources mandatory for course completion.

- Data show for power point presentations
- Up-to-date references in library
- Computers
- Internet connection

Course coordinator: **Dr. Salwa Ali Abou Elez Gawish**

Head of the department: **Dr. Salwa Gawish**

Date: