

# Philadelphia University Faculty of Science Department of Biotechnology and Genetic Engineering Winter Semester, 2009/2010

# Course Syllabus

Course Title: Human Genetics	Course code: 240234
Course Level: Third year	Course prerequisite (s) and/or corequisite (s):
Course Level. Third year	Basic Genetics (240231)
Lecture Time:	Credit hours: ?
S-T 8:10-9:00	

Academic Staff Specifics				
Name	Rank	Office No.	Office Hours	E-mail Address
Dr. Fayez Hamam	Assistant Prof.	823 (ext. 2345)	10:00-11:00 Sun and Tue Mon & Wed 10:.00-11:15	fhamam@philadelphia.edu.jo

### **Course module description:**

This module is advanced course in genetics it is a major requirements for the department. It is based on lectures and it focuses on the genetic basis of human diseases using interactive and analysis procedures.

### Course module objectives:

Explain the hereditary basis of human genetic diseases.

\* Understanding of the structure, function, and transmission of genes, the interactions both among genes and between genes and the environment, and the role of genetic factors in health and diseases

\* Demonstrate the ability to apply knowledge of hereditary disorders and congenital abnormalities to formulate appropriate diagnostic evaluation and patient management plans and to communicate information regarding genetic conditions to individuals of differing educational, socioeconomic, ethnic, and cultural backgrounds

### Course/module components:

Author(s)/Editor(s): Peter Sudbery Pearson Education Limited ISBN: second edition, 2002

## **Teaching methods:**

- Lectures, discussion groups, tutorials, problem solving, debates, etc.

- The use of Power Point presentation, Illustration with models, educational animations, and movies.

### **Learning outcomes:**

## • Knowledge and understanding

- Understanding of the structure, function, and transmission of genes, the interactions both among genes and between genes and the environment, and the role of genetic factors in health and diseases
- Demonstrate the ability to apply knowledge of genetic disorders and congenital anomalies to create suitable diagnostic assessment and patient management plans and to communicate information regarding genetic conditions to individuals of conflicting educational, socioeconomic, ethnic, and cultural backgrounds

## • Cognitive skills (thinking and analysis).

- The students will learn the ability to link between different problems and problem solving capabilities such as biostatistics
- The thinking skills will be developed by cheering students to conclude answers to different questions that the instructor intends to use during the presentation of the scientific material.
- The instructor intends to inspire the student's analytical thinking side via connections with general aspects in daily life or through questions, net searching, and home works.

## • Communication skills (personal and academic).

- Gain Teamwork skills
- The students have the option to share open discussion and to inquire questions during the class or any other times.
- Students have the chance to correspond with others especially professors, while probing answers for home works or through encouraging them to attend different scientific actions that are available in the department.
- Practical and subject specific skills (Transferable Skills).
  - Improve the ability to search using the scientific ways to get the concepts in human genetics.
  - Improve the ability to analyze different terms or phrases to its basic parts.

Allocation of Marks		
Assessment Instruments	Mark	
First examination	15%	
Second examination	15%	
Final examination: 50 marks	50%	
Reports, research projects, Quizzes, Home works.	20%	
Total	100%	

# Course/module academic calendar:

Week	Chapter	Basic and support material to be covered homework/rep	Pages
		and their due	
(1), (2) and	1	Introduction to human genetics and human genetic diseases	1-25
(3)		• Frequency and types of genetic disease	
		• Single-gene disorders	
		<ul> <li>Complexity in single-gene disorders</li> </ul>	
		Autosomal recessive	
		Autosomal dominant	
		• Sex-linked	
		Multifactorial or complex disorders	
		• Evidence for genetic factors in common diseases	
		• Genetic influences on personality disorders and pheno	
		traits	
		Chromosomal imbalances	
		Mitochondrial disorders	
		The Human Genome Project	
		Oniz #1 $(4\%)$	
(A) (5) and	2	Introduction to the structure of the human genome	27-51
(4), (5) and $(6)$	2	Amount of DNA in the human genome	27-31
(0)		Genes	
		Gene expression	
		Gene families	
		Dene families	
		<ul> <li>Fisculogenes</li> <li>Tondom report arrows of rDNA tDNA and history games</li> </ul>	
		<ul> <li>Tandeni repeat arrays of TKINA, tKINA and historie genes</li> <li>Intermediate repeated DNA</li> </ul>	
		Intermediate repeated DNA	
		• LINES: L1 and retrotransposition	
		• SINES: Alu family	
		• Processed pseudogenes	
		• Semsn DNA	
		Highly repetitive DNA	
		• Telomeres	
		• Tandem repeat arrays at centromeres	
		• Minisatellites	
		• Microsatellites	
		Human karyotype	
		Packaging DNA into chromosomes	
		Nucleosomes	
		• 30-nm fibre	
		• /00-nm fibre	
		Second quiz (4%)	
		First hour exam (15%)	
(7) + (8)	3	Structure & Mapping of the human genome (HG)	52-86
		<ul> <li>Importance of genomic maps</li> </ul>	
		<ul> <li>Sequence tagged sites</li> </ul>	
		Genetic maps	
		Physical maps	
		• Expression maps	
		• Integration of genetic, physical and expression maps	
		Second hour exam (15%)	

(9) + (10)	4	The IHGSC & Celera shotgun sequencing	
		4.1 introduction pages 87-89	
		4.2 Basic technology for seq. HG pages 89-92	
		4.2.3 shotgun seq. pages 92-94	
		4.3 IHGSC pages 94-98	
		4.4 Celera Genomics Strategy pages 98 -100	
		4.5 Single nucleotide polymorphisms pages 100-101	
		4.8 analysis of HG pages 104-107	
		4.8.2 Genes pages 113-114	
		4.8.4 How many genes? Pages115-116	
		4.5 Single nucleotide polymorphisms (SNP)	
		4.6 Accessing the data	
		4.7 Model Organisms	
		4.8 Analysis of HG Seq.	
		4.8.1.2 CpG	
		4.8.2 GENES	
		4.9 Exploiting HG Sequence	
		4.9.1.1 Identifying DNA seq. expressed in mRNA	
		4.9.2 Expressing profiling	
		4.9.3 Detecting sequence variation	
		Quiz #3 (4%)	
(11) + (12)	5	Single gene disorders/ Complex genetic diseases	1 40 1 41
		Introduction	140-141
		Cloning disease gene	141-146
		• Cystic fibrosis (CF)	14/-152
		• DMD	152-154
		Trinucleotide repeat expansion	154-158
		Trinucleotide repeat expansion	104-172
		Quiz # 4 (4%)	
(13) + (14)	6	Identifying genes of complex diseases	
		environmental effect	
		• Introduction	181-182
		• Evidence of genetic in complex diseases	183-184
		The composition of complex diseases	184-194
		<ul> <li>Identifying genes of complex diseases</li> </ul>	194-206
		<ul> <li>Genetic susceptibility to Alzheimer's disease</li> </ul>	216-220
(15)	7	Introduction to Gene therapy & Gene ther	225-246
(10)	,	models	220 210
l			
(16)		Final Evam	
(10)		F 11141 E2A4111	

## Expected workload:

On average students need to spend 2 hours of study and preparation for each 50-minute lecture/tutorial.

### **Attendance policy:**

Absence from lectures and/or tutorials shall not go beyond 15%. Students who exceed the 15% boundary without a medical or emergency reason acceptable to and endorsed by the Dean of the relevant college/faculty shall not be permitted to take the final examination and shall receive a mark of zero for the course. If the excuse is approved by the Dean, the student shall be considered to have withdrawn from the course.