University of Nicosia, Cyprus

Course Code	**Course Title**	**ECTS Credits**
BIOL-232	Human Molecular Genetics	6
Department	**Semester**	**Prerequisites**
Life and Health Sciences	Fall	BIOL-101 ,-102 General Biology I, II
Type of Course	**Field**	**Language of Instruction**
Required	Genetics	English
Level of Course	**Year of Study**	**Lecturer**
1st Cycle	2nd	Dr. Evi Farazi
Mode of Delivery	**Work Placement**	**Co-requisitesrs**
face-to-face	N/A	None

Objectives of the Course:

This course aims to promote students' comprehension of genetic mutations, how chromosomes and genes are related in health and disease, and how comparative analysis data of genomes is used to study diseases and the evolutionary origin of human DNA. The main objectives of the course are to:

- Review the classic Mendelian inheritance patterns and analysis and discuss in depth non-Mendelian inheritance.
- Introduce students to the concept of population genetics and the molecular methods used in genetic research.
- Use examples from scientific literature to demonstrate how genetic mapping and LOD score analysis are used in the identification of human disease genes.
- Use examples to examine the role of DNA mutations and its association with genomic instability, diseases and cancer.
- Review key scientific papers and make students aware of the latest technological and scientific discoveries in the field of genetics.

Learning Outcomes:

After completion of the course students are expected to be able to:

1. Analyze human pedigrees and determine the mode of inheritance for any type of disease.
2. Recommend the appropriate molecular method to address a genetic problem experimentally.

3. Calculate gene distances and map genes based on recombination frequencies; and linkage status using LOD score analysis.
4. Determine allelic, genotypic, and phenotypic frequencies from population data, and the number of genes involved in polygenic traits.
5. Define mutation and classify the various types of mutations and discuss their role in genetic diseases using examples from published papers.
6. Design an experimental study in order to identify a gene and to characterize its function in the development of a genetic disease.
7. Discuss the basis of modern molecular methods used in genetic testing.

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Course Contents:

1. Mendelian analysis & basic human pedigree analysis
2. Chromosomal basis of inheritance, sex linkage, X- & Y-linked inheritance in humans
3. Human chromosomes, structure, analysis, and abnormalities
4. DNA structure & gene expression
5. Extensions of Mendelian analysis & population genetics
6. Molecular methods used to analyze DNA & gene expression
7. The human genome project
8. Organization of the human genome
9. Human gene expression
10. Mutation, DNA repair, & genomic instability
11. Gene evolution, comparative genomics & evolution of human populations
12. Genetic mapping of Mendelian characters
13. Identification of genes causing human disease
14. Mapping & identification of genes of complex human diseases
15. Loss & gain of function mutations
16. Cancer genetics
17. Genetic testing
18. Genetic manipulation of cells & animals
19. Novel approaches to studying genetics (functional genomics, proteomics, & bioinformatics)

Learning Activities and Teaching Methods:

Lectures 3h/week and tutorial exercises 1h/week (tutorials involve genetic problem solving, reading and discussion of scientific literature in the field of genetics, discussion of novel technological developments in the field of genetics); Cooperative learning activities.

Assessment Methods:

Assignments, (Problem Sets), Tests and Mid-term Exam; Final Exam

Required Textbooks/Reading:

Authors	Title	Publisher	Year	ISBN
Klug, Cummings, Spencer, Palladino	Concepts of Genetics	Pearson International Edition	9th edition	978-0-321-54098-0

Recommended Textbooks/Reading:

Authors	Title	Publisher	Year	ISBN
Tom Strachan & Andrew Read	Human Molecular Genetics 3	Garland Science (Taylor & Francis group)	2004, 3rd Edition	0-8153-4182-2