UNIVERSITY of NICOSIA

Course Syllabus

Course Code	Course Title	ECTS Credits
BIOL-232	Human Molecular Genetics	6
Prerequisites	**Department**	**Semester**
BIOL-101 BIOL-102	Life Sciences	Fall
Type of Course	**Field**	**Language of Instruction**
Required	Human Biology	English
Level of Course	**Lecturer(s)**	**Year of Study**
1st Cycle	Dr Maria Christofidou	2nd
Mode of Delivery	**Work Placement**	**Corequisites**
Conventional	N/A	None

Course Objectives:

The purpose of this course is to present the basic principles of human molecular genetics and how these principles can be applied in clinical practice. The course will cover an array of topics, including the structure and function of the human genome, the types of genetic mutations, the patterns and analysis of Mendelian inheritance, the molecular and chromosomal basis of inherited diseases, e.g. cancer and other diseases with a genetic component. The students will be introduced to the latest methodology of analysis of genetic diseases and to the contribution of genome projects towards human health. The course will also cover the fields of pharmacogenomics, personalised medicine and gene therapy. There will also be an introduction to genetic diagnosis, genetic counselling through the analysis of pedigrees and clinical scenarios.

Learning Outcomes:

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

1. Understand the structure and organisation of the human genome, the genetic variation, the modes of inheritance and the interaction between genes and environment
2. Understand the molecular aetiology of common, rare and multifactorial genetic diseases
3. Apply in practice their knowledge about genetic diagnosis and clinical genetics: collecting a family history, drawing human pedigrees, providing genetic counselling, recommending prenatal screening and testing when it is appropriate

4. Know the different genome projects and become aware of the novel discoveries in diagnosis, prognosis and treatment of genetic diseases

Course Content:

Topics covered during the course include:

1. Structure and organization of the human genome: DNA and chromosomes
2. Genome organization and expression
3. Genomics
4. Numerical and structural chromosomal abnormalities
5. Monogenic diseases
6. Modes of human inheritance
7. Multifactorial inheritance and complex diseases
8. Mitochondrial inheritance
9. Epigenetic inheritance and parental imprinting
10. Mutations and repair mechanisms
11. Genetic polymorphisms
12. Animal models of human genetic diseases
13. Human pedigrees
14. Cancer genetics
15. Genetic testing and guidance
16. Basic principles of gene therapy
17. Clinical case studies/scenarios of molecular genetics in clinical genetics (prenatal diagnosis and screening, genetic counselling, disease prognosis)

Learning Activities and Teaching Methods:

Lectures, in-class examples, discussion of possible clinical scenarios, audio-visual learning material, class discussions

Assessment Methods:

Coursework and Assignments, Midterm exam, Final exam

Recommended Textbooks / Reading:

*** Students are advised to read the relevant reading material provided with each lecture as seen on the course page on Moodle ***

Title	Author(s)	Publisher	Year	ISBN
Human Molecular Genetics	Strachan Tom, Read Andrew	Garland Science	2018	9780815345893
Genetics and Genomics in Medicine	Strachan Tom, Lucassen Anneke	Garland Science	2022	9780367490812
Essentials of Genetics	William S Klug, Michael Cummings, Charlotte A. Spencer, Michael A Palladino, Darrell Killian	Pearson	2019	ISBN-10: 0134898419 ISBN-13: 978-0134898414
Or the latest edition of the above mentioned books. Updated scientific articles (primary and secondary) based on the lecture material				