

COURSE OUTLINE

(1) GENERAL

SCHOOL	School of Applied Biology and Biotechnology		
ACADEMIC UNIT	Biotechnology		
LEVEL OF STUDIES	Undergraduate		
COURSE CODE	3607	SEMESTER	9th
COURSE TITLE	Genetics of Human Diseases		
INDEPENDENT TEACHING ACTIVITIES <i>if credits are awarded for separate components of the course, e.g. lectures, laboratory exercises, etc. If the credits are awarded for the whole of the course, give the weekly teaching hours and the total credits</i>		WEEKLY TEACHING HOURS	CREDITS
Lectures		2,3	0,16
Laboratory Exercises		2,3	0,09
Assignments		1,5	0,06
Total			5,00
<i>Add rows if necessary. The organisation of teaching and the teaching methods used are described in detail at (d).</i>			
COURSE TYPE <i>general background, special background, specialised general knowledge, skills development</i>	General background		
PREREQUISITE COURSES:	None Required		
LANGUAGE OF INSTRUCTION and EXAMINATIONS:	Greek		
IS THE COURSE OFFERED TO ERASMUS STUDENTS	Yes		
COURSE WEBSITE (URL)	https://mediasrv.aua.gr/eclass/courses/281/		

(2) LEARNING OUTCOMES

<p>Learning outcomes <i>The course learning outcomes, specific knowledge, skills and competences of an appropriate level, which the students will acquire with the successful completion of the course are described.</i></p> <p><i>Consult Appendix A</i></p> <ul style="list-style-type: none"> • <i>Description of the level of learning outcomes for each qualifications cycle, according to the Qualifications Framework of the European Higher Education Area</i> • <i>Descriptors for Levels 6, 7 & 8 of the European Qualifications Framework for Lifelong Learning and Appendix B</i> • <i>Guidelines for writing Learning Outcomes</i>
<p>This is a basic course that presents the principles of Genetic Diseases. It aims to introduce students to the basic approaches for understanding the Genetics of inherited diseases, genetic diagnostics and modern therapeutic approaches. It also reports extensively to the genetic, molecular and chromosomal basis of monogenic inherited diseases, diseases with complex inheritance, and cancer. Finally, the aim of the course is the understanding of the importance of basic principles of genetic diseases as a basis for application of diagnosis, personalized medicine, and novel treatments.</p> <p>Upon successful completion of this course the student will be able to:</p> <ul style="list-style-type: none"> • Understand the basic concepts of Genetics. • Provide insight into the genetic and molecular basis of monogenic and complex diseases and cancer. • Understand the basic pathogenetic mechanisms of inherited diseases and gain knowledge about diagnostic methods and innovative therapeutic approaches.

- Have extensive knowledge of the basic concepts and applications of pharmacogenetics and pharmacogenomics and how individual variations affect personalized treatment.
- Have acquired knowledge for risk assessment in genetic diseases and determining recurrence risk.
- Have an understanding of the ethical dilemmas that arise in medical genetics and is able to approach them through a scientific background.

Upon successful completion of the lab the student:

- Has acquired knowledge of clinical cases of human genetics diseases.
- Has a deeper understanding of the applications of genetics and how it dictates the medical approach to be followed.
- Is able to approach genetic diseases in terms of their genetic profile, molecular and medical diagnosis, and clinical treatment, treatment and outcome.
- Has learned about the stress system and the generalized syndrome of resistance in glucocorticoids, infantile systemic hyalinosis, genetic and endocrine components in sex disorders, the role of epigenetics in childhood abuse and the genetics of cancer.
- Understands the genetic panels in cancer as well as STR analysis in forensics.

General Competences

Taking into consideration the general competences that the degree-holder must acquire (as these appear in the Diploma Supplement and appear below), at which of the following does the course aim?

Search for, analysis and synthesis of data and information, with the use of the necessary technology
Adapting to new situations
Decision-making
Working independently
Team work
Working in an international environment
Working in an interdisciplinary environment
Production of new research ideas

Project planning and management
Respect for difference and multiculturalism
Respect for the natural environment
Showing social, professional and ethical responsibility and sensitivity to gender issues
Criticism and self-criticism
Production of free, creative and inductive thinking
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Others...
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- Retrieve, analyze and synthesize data and information, with the use of necessary technologies.
- Adaptation to new situations.
- Decision making.
- Work autonomously.
- Work in teams.
- Creation of novel research ideas.

(3) SYLLABUS

- Introduction to disease genetics
- The human genome and the chromosomal basis of heredity
- The Human Genome: Gene Structure and Function
- Tools for human molecular genetics
- Principles of clinical cytogenetics
- Clinical cytogenetics: Disorders of the autosomes and sex chromosomes
- Patterns of single-gene inheritance
- Complex inheritance of common multifactorial disorders
- Genetic variation in populations: mutations and polymorphism
- Mapping and identifying human genes related to diseases
- Hemoglobinopathy as standards of molecular disease
- Molecular, Biochemical, and Cellular Basis of Genetic Disease
- The treatment of genetic diseases

- Developmental genetics and birth defects
- Prenatal diagnosis
- Cancer genetics and genomics
- Personalized medical genetics
- Pharmacogenetics and Pharmacogenomics
- Genetic counselling and risk assessment
- Ethical issues and medical genetics

(4) TEACHING and LEARNING METHODS - EVALUATION

DELIVERY <i>Face-to-face, Distance learning, etc.</i>	Face-to-face & synchronous and asynchronous learning online	
USE OF INFORMATION AND COMMUNICATIONS TECHNOLOGY <i>Use of ICT in teaching, laboratory education, communication with students</i>	Use of the e-learning Moodle system, with uploaded notes, lectures in videos, exercises for practice and communication with students.	
TEACHING METHODS <i>The manner and methods of teaching are described in detail. Lectures, seminars, laboratory practice, fieldwork, study and analysis of bibliography, tutorials, placements, clinical practice, art workshop, interactive teaching, educational visits, project, essay writing, artistic creativity, etc.</i> <i>The student's study hours for each learning activity are given as well as the hours of non-directed study according to the principles of the ECTS</i>	Activity	Semester workload
	Lectures	30
	Laboratory Exercises	30
	Assignment	20
	Homework	45
	Course total	125
STUDENT PERFORMANCE EVALUATION <i>Description of the evaluation procedure</i> <i>Language of evaluation, methods of evaluation, summative or conclusive, multiple choice questionnaires, short-answer questions, open-ended questions, problem solving, written work, essay/report, oral examination, public presentation, laboratory work, clinical examination of patient, art interpretation, other</i> <i>Specifically-defined evaluation criteria are given, and if and where they are accessible to students.</i>	<p>I. Theory (50%): Written final exam including:</p> <ul style="list-style-type: none"> - Multiple choice questions - Judgement Questions <p>II. Laboratory: Written final exam (30%) including:</p> <ul style="list-style-type: none"> - Multiple choice questions - Judgement Questions - Problem Solving <p>Assignment Presentation (20%)</p>	

(5) ATTACHED BIBLIOGRAPHY

Title	MEDICAL GENETICS (7 th Ed.)
Author	THOMSON & THOMSON
ISBN	9789604890620
Publisher	Broken Hill Publishers Ltd. Paschalidis Medical Publications
Year	2011