

COURSE OUTLINE

(1) GENERAL

SCHOOL	School of Applied Biology and Biotechnology		
ACADEMIC UNIT	Biotechnology		
LEVEL OF STUDIES	Undergraduate		
COURSE CODE		SEMESTER	9
COURSE TITLE	Computational Translational Genetics		
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		2,0	0,19
Laboratory Exercises		2,0	0,12
Total			4,00
<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)	http://gbt.aua.gr/proptixiaka2.html		

(2) LEARNING OUTCOMES

<p>Learning outcomes</p> <p><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>The course presents the fundamental principles of translational genetics and the computational methods applied to exploit data provided by state-of-the-art technologies (eg Whole Genome Sequencing, RNA-seq, DNA methylation assay). The purpose is to acquaint the student with computational techniques for the combined analysis of genetic information at the sequence level as well as the transcriptional and translational profile and epigenetic modifications, and the discovery of correlations with phenotypic characteristics. The course material covers a wide range of modern computational approaches, such as translational bioinformatics, machine learning and deep learning techniques, and computational models for structure prediction and interaction networks prediction.</p> <p>The course focuses on methodologies for the analysis of genetic diversity and polymorphisms at the population level in terms of specific phenotypic traits through genome-wide association studies and the calculation of the polygenic risk index, as well as modern approaches for the computational analysis of genetic diversity in</p>

the context of genetic epidemiology. It focuses on the student's expertise in computational tools and methods for the clinical utility of electronic health records and medical data and deals with the application of artificial intelligence and machine learning algorithms to investigate genotype-phenotype associations. Finally, it includes a more general overview of the architecture of computational applications for the clinical diagnosis, prevention and treatment of pathologies based on the clinical and genetic profile.

Upon successful completion of the course, the student will be able to:

- Understand the basic concepts of translational genetics.
- Be familiar with genome wide association studies, the concept of genetic linkage and haplotype investigation and is able to apply computational methods for phenotype-genotype correlation and determination of polygenic risk score.
- Have extensive knowledge of the genome organization as well as of the genetic interaction networks and their visualization methods.
- Understand the basic mechanisms of epigenetics and be familiar with the application of basic methods of searching, analyzing and predicting epigenetic modifications.
- Understand the basic principles of the architecture of computational genetics applications.
- Be familiar with the concepts of disease prevention and treatment and with the development, management, and utilization of electronic medical data.
- Be familiar with the use of computational tools for data retrieval and the design of a comprehensive pipeline for therapeutic strategies.
- Monitor the developments, understand the modern methodologies and evaluate the research results that arise in the field of translational genetics.

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
.....
Others...
.....*

- 1) Retrieve, analyze and synthesize data and information, with the use of necessary technologies.
- 2) Adaptation to new situations.
- 3) Decision making.
- 4) Work autonomously.
- 5) Work in teams.
- 6) Creation of novel research ideas.
- 7) Design and implementation of research projects.

(3) SYLLABUS

- Introduction to computational translational genetics.

- Genome-Wide Association Studies (GWAS).
- Distribution of alleles in populations, genetic linkage and haplotype estimation.
- Methods of genotyping analysis to identify point mutations and structural variants.
Polygenic risk score (PRS).
Computational methods for genotype prediction based on Single Nucleotide Polymorphisms (SNP).
- Computational genetic epidemiology.
- Mathematical models for genome organization.
- Gene interaction analysis for quantitative traits.
- Computational prediction of epistatic interactions.
- Genetic networks, graph theory and visualization.
- Computational models for predicting epigenetic mechanisms, gene expression from histone modification.
- Detection and analysis of DNA methylation patterns.
- DNA damage analysis algorithms.
- Prediction of post-translational modifications of proteins.
- Machine learning and deep learning techniques in computational genetics.
- Disease prevention, treatment and management through computational translational genetics.
- Electronic health records and medical data.
- Architecture of online genetics applications.

(4) TEACHING and LEARNING METHODS - EVALUATION

DELIVERY <i>Face-to-face, Distance learning, etc.</i>	Face-to-face		
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, 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. 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	40	
	Laboratory Exercises	30	
	Homework	30	
	Course total	100	
STUDENT PERFORMANCE EVALUATION <i>Description of the evaluation procedure 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. Theory: Written final exam including: <ul style="list-style-type: none"> - Multiple choice questions - Judgement Questions II. Laboratory: Written final exam including:		

Specifically-defined evaluation criteria are given, and if and where they are accessible to students.

- Multiple choice questions
- Judgement Questions

(5) ATTACHED BIBLIOGRAPHY

Title Medical Informatics
Author NIKIFORIDIS CH. GEORGIOS
ISBN 978-960-372127
Publisher LITSAS K & SIA E.E. Printing & Publishing Company
Year 2009

Title Medical Genetics
Author GEORGE SACK
ISBN 9789603941415
Publisher PARISIANOU S.A.
Year 2002