

## COURSE OUTLINE

### (1) GENERAL

<b>SCHOOL</b>	School of Applied Biology and Biotechnology	
<b>ACADEMIC UNIT</b>	Biotechnology	
<b>LEVEL OF STUDIES</b>	Undergraduate	
<b>COURSE CODE</b>	<b>279</b>	<b>SEMESTER</b> 8
<b>COURSE TITLE</b>	Data Analysis in Genetics	
<b>INDEPENDENT TEACHING ACTIVITIES</b> <i>if credits are awarded for separate</i>	<b>WEEKLY TEACHING HOURS</b>	<b>CREDITS</b>
Lectures and Practicals	3	1.56
Laboratory exercises	2	0.56
Group or individual laboratory		0.56
Independent study		2.32
	<b>Total</b>	<b>5</b>
<i>Add rows if necessary. The organisation of teaching and the teaching methods</i>		
<b>COURSE TYPE</b> <i>general background, special background, specialised general knowledge, skills development</i>	Field of Science	
<b>PREREQUISITE COURSES:</b>		
<b>LANGUAGE OF INSTRUCTION and EXAMINATIONS:</b>	Greek	
<b>IS THE COURSE OFFERED TO ERASMUS STUDENTS</b>	No	
<b>COURSE WEBSITE (URL)</b>	<a href="https://oeclass.aua.gr/eclass/modules/auth/opencourses.php?fc=37">https://oeclass.aua.gr/eclass/modules/auth/opencourses.php?fc=37</a>	

### (2) LEARNING OUTCOMES

#### Learning outcomes

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.

Consult Appendix A

- Description of the level of learning outcomes for each qualifications cycle, according to the Qualifications Framework of the European Higher Education Area
- Descriptors for Levels 6, 7 & 8 of the European Qualifications Framework for Lifelong Learning and Appendix B
- Guidelines for writing Learning Outcomes

The course is designed to introduce Next Generation Sequencing methods and Data Analysis Tools. Emphasis will be put on the understanding and utilization of these concepts and algorithms. The objective is to help the students to reach rapidly the frontier of NGS big data analysis and be able to use standard and deep learning tools to solve problems in an interdisciplinary environment.

After completing this course, the student will be able to:

- explain the paradigm shift in Biology and the need for Big Data Analysis
- understand sequencing technologies: chain termination method, sequencing by synthesis, Single Molecule Real Time sequencing
- describe the structure of FASTQ, SAM, VCF, GFF3, BED files
- explain de novo and reference based assembly

- describe types of genomic variants and how they are determined
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- describe structural and functional annotation results
- understand experimental and RNA sequencing design
- understand differential gene expression
- explain metagenomic sequencing, assembly and classification
- understand how to visualize, compare and analyze genomics data
- describe proteomics techniques and data analysis results
- explain the biological interpretation of metabolomics data
- analyze data to determine genetic variation within populations

After completing the lab, the student will be able to use computational tools for:

- Quality analysis of next generation sequencing data
- RNA sequencing data mapping on reference genome
- Variant calling
- Differential gene expression
- Proteomics data analysis
- Genetic data analysis to detect polymorphisms associated with autoimmune diseases

### 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...*  
 .....

Search for, analysis and synthesis of data and information, with the use of the necessary technology  
 Working independently  
 Working in an interdisciplinary environment

### (3) SYLLABUS

Introduction  
 DNA Sequencing Technology  
 Genome Assembly  
 Variant Calling  
 Genome Annotation  
 RNA Sequencing  
 Metagenomics  
 Genome Data Visualization  
 Proteomics  
 Metabolomics  
 Human DNA polymorphisms

### (4) TEACHING and LEARNING METHODS - EVALUATION

<b>DELIVERY</b> <i>Face-to-face, Distance learning, etc.</i>	in the classroom	
<b>USE OF INFORMATION AND COMMUNICATIONS TECHNOLOGY</b> <i>Use of ICT in teaching, laboratory education, communication with students</i>	Lectures: Power point presentations Labs: Use of biological databases and bioinformatics software	
<b>TEACHING METHODS</b> <i>The manner and methods of teaching are</i>	<b>Activity</b>	<b>Semester workload</b>
	Lectures	39 hr (1.56 ECTS)

<i>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>	Laboratory work	14 hr (0.56 ETCS)
	Written assignments	14 hr (0.56 ECTS)
	Autonomous study	58 hr (2.32 ECTS)
	Course total	125 hr (5 ECTS)
<p align="center"><b>STUDENT PERFORMANCE EVALUATION</b></p> <p><i>Description of the evaluation procedure</i></p> <p><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></p> <p><i>Specifically-defined evaluation criteria are given, and if and where they are accessible to students.</i></p>	<p>I. Theory: Written Examination (50%) II. Lab: Written Examination (35%) Written assignments (15%)</p>	

### (5) ATTACHED BIBLIOGRAPHY

<p>1. Εργαστηριακοί Υπολογισμοί στις Βιολογικές Επιστήμες (ΑΚΑΔΗΜΑΪΚΕΣ ΕΚΔΟΣΕΙΣ Ι. ΜΠΑΣΔΡΑ &amp; ΣΙΑ)</p> <p>2. Βιοπληροφορική &amp; Λειτουργική Γονιδιωματική (ΑΚΑΔΗΜΑΪΚΕΣ ΕΚΔΟΣΕΙΣ Ι. ΜΠΑΣΔΡΑ &amp; ΣΙΑ)</p>
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