Course Title	**Introduction to Bioinformatics**				
Course Code	ABS409				
Course Type	Program specific elective				
Level	BSc 1st level				
Year / Semester	4th / 7th Semester				
Teacher's Name	Dr Panayiotis Paoullis / Dr Chrystalla Iosif				
ECTS	6	Lectures / week	3	Laboratories / week	2
Course Objectives	Bioinformatics is the application of computational tools for the analysis and interpretation of biological data. It is an interdisciplinary field, which harnesses computer science, mathematics, and biology. Bioinformatics is widely accepted as a major future direction of biomedical sciences, beneficial to students. This course's main purpose is to provide students with the theoretical background and working knowledge of the techniques employed in bioinformatics, including learning the R language, to solve biological problems. The aim of this course is to: - Expose students to the multitude of the different bioinformatics software tools which are used to analyze large data sets. - Students will become knowledgeable and acquire the basic skills to use the most appropriate and suitable tools to solve biological questions - Students will become competent and work independently in applying bioinformatic pipelines to extract and analyze significant data and reach valid conclusions.				
Learning Outcomes	By the end of this course students will be able to: Understand the importance of Bioinformatics in Applied Biomedical Science. Learn and apply the use of the R language in bioinformstics analysis Understand genomics/transcriptomics/ proteomics and become familiar with the concept of homologs, orthologs and paralogs. Become familiar with DNA and protein databases/information retrieval and learn about sequence alignment. Understand gene annotation, identification of introns and exons and how promoters regulate gene expression. Learn about Phylogenetic trees and how to perform phylogenetic analysis. Learn about microarrays and the different types of omics, the generated data and how to apply bioinformatic algorithms for their analysis.				
Prerequisites	-		Required		-
Course Content	Introduction to bioinformatics, the process of DNA replication, transcription, translation, and basic genomics /transcriptomics/proteomics and learn the terms homologs, orthologs and paralogs.				

- Introduction and applications of the R computer language
- Introduction to biological databases, learning about DNA and protein databases and information retrieval. Compare sequences using Basic Local Alignment Search Tool (BLAST) and FASTA. Explore various biological Databases.
- Introduction to Sequence Alignment/ Distinguish between global and local alignment. Understand the basic statistics used in bioinformatics including scoring matrices, hidden Markov models (HMMs) and statistical significance of sequence alignment.
- Familiarize and perform pairwise sequence alignment and multiple sequence alignment.
- Gene and Promoter prediction/the importance of gene annotation, including detection of location of open reading frames and identification of introns and exons. Learn how promoters regulate gene expression and the different methods of promoters' identification, ab initio-based Algorithms, or expression profile-based methods.
- Phylogenetics: Define evolution and introduce the concept of phylogenetics. Introduction to Phylogenetic trees, their use and construction methods. Identify how to choose substitution models. Be able to perform phylogenetic analysis.
- Microarrays: Become familiar with the principles and applications of microarrays.
- Polymorphisms: Understand the basics of molecular evolution and the origins of polymorphisms. SNP detection methods.
- Genomics: Introduced to genome mapping, sequencing, assembly, annotation, functional genomic analysis and comparative genomic analysis. Gain knowledge of current sequencing technique
- Proteomics: Introduction to proteomics and the various proteomics techniques, including X-ray crystallography and nuclear magnetic resonance spectroscopy. Analysis of protein expression, posttranslational modifications, protein sorting and protein-protein interaction.
- Metabolomics: Understand the basis of metabolomics, methods used including mass spectrometry and NMR and data analysis.
- Predictive modelling: Understand the concept of predictive modelling and familiarized with the various predictive model algorithms including Random Forest, and LASSO, and cross validation methods, such as data randomization and LOOCV.
- Computational: Learn how to use various bioinformatics algorithms for performing different bioinformatics analysis using RStudio (R language), Python and Linux.

Theory:

2

	Molecular biology central dogma Introduction to bioinformatics biological databases Sequence Alignment Introduction to statistical methods Gene and Promoter prediction Phylogenetics Microarrays Polymorphism Genomics Proteomics Metabolomics Predictive modelling. Computer exercises/assignments: As part of the course, the student will have the opportunity to apply their theoretical knowledge. Introduction and use of RStudio and Linux. Retrieve information from different sequencing databases. Use BLAST and FASTA. Use algorithms to perform pairwise and multiple sequencing alignment. Multivariate analysis for "omics" data. Analysis of microarrays results. Use of bioinformatics tools for gene prediction, operon prediction, promoter prediction and gene annotation. Perform PCA analysis, use clustering algorithms, perform differential expression analysis and functional enrichment analysis. Perform pathway analysis and generate pathway networks. Protein structure visualization, comparison and classification. Construct phylogenetic trees based on biological sequence data. Predictive modelling exercises.
Teaching	Teaching methodology includes lectures on the theoretical background and computational exercises to better understand concepts of Bioinformatics. Detailed lecture notes are presented. Students are also introduced to specific databases. During lectures, discussions are carried out and students are encouraged to answer questions and draw their own conclusions. As part of developing students' skills, students will use a variety of bioinformatics tools. Appropriate preparation and demonstration precede each computational exercise. Assessment of bioinformatics tools usage includes the evaluation of analysis results reports submitted by each student after each exercise.
Bibliography	(a) Textbooks: - Practical Bioinformatics, 1st ed., Agostino, M., Garland Science, 2013 - Essential Bioinformatics, Xiong J., Cambridge university press, 2006 (b) References: - Bernstam, Elmer V., Jack W. Smith, and Todd R. Johnson. 2010. "What Is Biomedical Informatics?" Journal of Biomedical Informatics 43 (1): 104–10. - Bhardwaj, Kartik Krishna, Siddhant Banyal, and Deepak Kumar Sharma. 2019. "Artificial Intelligence Based Diagnostics, Therapeutics and Applications in Biomedical Engineering and Bioinformatics." In Internet of Things in Biomedical Engineering, 161–87. Elsevier. - Liu, Yuan, Yamei Chen, and Leng Han. 2023. "Bioinformatics: Advancing Biomedical Discovery and Innovation in the Era of Big Data and Artificial Intelligence." The Innovation Medicine 1 (1):

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- Luo, Jake, Min Wu, Deepika Gopukumar, and Yiqing Zhao. 2016. "Big Data Application in Biomedical Research and Health Care: A Literature Review." Biomedical Informatics Insights 8 (January): 1–10.
- Pereira, Rute, Jorge Oliveira, and Mário Sousa. 2020. "Bioinformatics and Computational Tools for Next-Generation Sequencing Analysis in Clinical Genetics." Journal of Clinical Medicine Research 9 (1).</td></tr>
<tr><td>Assessment</td><td>Course Work 40%
- Mid-term Test 20%
- Computational Exercises/Lab reports 20%
Final Exam 60%

For student evaluation, the overall grade is determined by a written midterm exam (20%), a laboratory grade (20%) and a written final exam (60%).

The mid-term exam is carried out between the 6th and 8th week, and it mainly includes short answer- questions and problem- solving questions and examines specific modules of the course.

As far as the laboratory grade is concerned, it comprises of the evaluation of the laboratory reports (60% of the laboratory grade) submitted by the students after every experiment and a final laboratory examination (40% of the laboratory grade) which mainly includes short answer questions and problem-solving questions. In their laboratory reports, students are asked to describe the experimental procedure, to evaluate and analyse their results and to answer specific questions. The following criteria are taken into account when evaluating laboratory reports: (a) experimental data collection (30%), (b) data analysis (40%), and application of theory to draw conclusions (30%).

The final exam of the course is carried out during the 14th-16th week of each semester and includes short answer questions, decision questions, and problem-solving questions regarding all course modules.

The final assessment of the students is formative and summative and is assured to comply with the subject's expected learning outcomes and the quality of the course.</td></tr>
<tr><td>Language</td><td>Greek, English</td></tr>
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