

## SYLLABUS

### 1. Information regarding the programme

1.1 Higher education institution	<b>Babeş Bolyai University</b>
1.2 Faculty	<b>Faculty of Biology and Geology</b>
1.3 Department	<b>Department of Molecular Biology and Biotechnology</b>
1.4 Field of study	<b>Biology</b>
1.5 Study cycle	<b>Master</b>
1.6 Study programme / Qualification	<b>Bioinformatics applied in life sciences</b>

### 2. Information regarding the discipline

2.1 Name of the discipline (en) (ro)	<b>Applied Genomics in human health</b> <b>Genomică aplicată în sănătatea umană</b>						
2.2 Course coordinator	<b>Rareş Călin Lucaciu</b> <b>Sef lucr. Dr. Cruceriu Daniel</b>						
2.3 Seminar coordinator	<b>Rareş Călin Lucaciu</b> <b>Sef lucr. Dr. Cruceriu Daniel</b>						
2.4. Year of study	<b>2</b>	2.5 Semester	<b>3</b>	2.6. Type of evaluation	<b>E</b>	2.7 Type of discipline	<b>Elective</b>
2.8 Code of the discipline	<b>BME1132</b>						

### 3. Total estimated time (hours/semester of didactic activities)

3.1 Hours per week	<b>4</b>	Of which: 3.2 course	<b>2</b>	3.3 seminar/laboratory	<b>2</b>
3.4 Total hours in the curriculum	<b>56</b>	Of which: 3.5 course	<b>28</b>	3.6 seminar/laboratory	<b>28</b>
Time allotment:	hours				
Learning using manual, course support, bibliography, course notes	24				
Additional documentation (in libraries, on electronic platforms, field documentation)	18				
Preparation for seminars/labs, homework, papers, portfolios and essays	16				
Tutorship	8				
Evaluations	4				
Other activities: .....					
3.7 Total individual study hours	70				
3.8 Total hours per semester	126				
3.9 Number of ECTS credits	5				

### 4. Prerequisites (if necessary)

4.1. curriculum	<ul style="list-style-type: none"> <li>• Cell and molecular biology</li> <li>• Genetics, genomics and functional genomics</li> <li>• Biostatistics</li> </ul>
4.2. competencies	<ul style="list-style-type: none"> <li>• Interpretation of cell and molecular biology data</li> <li>• Beginner programming skills (bash and R)</li> </ul>

### 5. Conditions (if necessary)

5.1. for the course	<ul style="list-style-type: none"> <li>• Video projector (for on-site activities)/ MS Teams or ZOOM online platforms</li> <li>• Blackboard (on site)/ graphic pad (for online communication)</li> </ul>
5.2. for the seminar /lab activities	<ul style="list-style-type: none"> <li>• Video projector (for on-site activities)/ MS Teams or ZOOM online platforms</li> <li>• Blackboard (on site)/ graphic pad (for online communication)</li> </ul>

	<ul style="list-style-type: none"> <li>• PC desktops or notebooks (at least 1 unit per 3 users or, alternatively one unit per each student if online activities are planned)</li> <li>• Attendance of a minimum 90% of practical work / seminar is prerequisite for admission at written exam</li> </ul>
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## 6. Specific competencies acquired

<b>Professional competencies</b>	<p>By completing this course, students will be able to</p> <ul style="list-style-type: none"> <li>• Understand the concept of sequencing and the principles behind using OMICs data, including DNA-seq, RNA-seq, and Single Cell analysis.</li> <li>• Understand the principles of genome/transcriptome assembly using algorithms like “de Bruijn graph” and be able to use specific bioinformatics software such as Trinity and Spades.</li> <li>• Understand Single Nucleotide Polymorphisms (SNPs), Indels (Insertions and Deletions), Copy Number Variations (CNVs), and generate a DNA data analysis pipeline to detect genomic variations. This pipeline includes steps like Quality Control (QC), Mapping, Mapping QC, Variant Detection, and Annotation.</li> <li>• Understand RNA data concepts and generate an analysis pipeline that includes QC, RNA mapping, quantification (FPKM, RPKM, TPM, TMM), differential gene expression analysis (DEG analysis), and pathway analysis.</li> <li>• Understand the concept of Single Cell analysis and its applications in biomedical research.</li> <li>• Understand microbiome sequencing, including 16S amplicon sequencing and metagenomics.</li> <li>• Apply theoretical knowledge to solve practical problems in genomic and transcriptomic research related to human health.</li> <li>• Be familiar with and use various software tools for analyzing OMICs data, such as MaSuRCA for assembly and other specific tools for genomic variation and transcriptomic analysis.</li> </ul>
<b>Transversal compt</b>	<ul style="list-style-type: none"> <li>• To use theoretical concepts in solving practical problems in the fields of genomics and transcriptomics in human health.</li> <li>• To understand the concept of gene signatures and drug discovery.</li> <li>• To facilitate the transfer of information for understanding the human genome in general and pathogenomics in particular, by acquiring and applying knowledge from related fields such as cytology, genetics, molecular biology, biostatistics, and bioinformatics."</li> </ul>

## 7. Objectives of the discipline (outcome of the acquired competencies)

7.1 General objective of the discipline	<ul style="list-style-type: none"> <li>• Knowledge of the main human genome sequencing technologies and the methods for interpreting the raw data obtained through them.</li> </ul>
7.2 Specific objective of the discipline	<ul style="list-style-type: none"> <li>• To explain the operating principles of various widely used sequencing technologies.</li> <li>• To analyze OMICs data from genome and transcriptome assembly to variant identification and gene expression analysis.</li> <li>• To interpret the results obtained in a clinical context.</li> </ul>

## 8. Content

8.1 Course	Teaching methods	Remarks
1. <b>Introduction</b> to the Applied Genomics in Human Health course: syllabus and educational objectives.	<ul style="list-style-type: none"> <li>• Interactive exposure</li> <li>• Presentation</li> <li>• Explanation</li> </ul>	
2. <b>Sequencing:</b> introduction to the concept, operation of a sequencing machine, and detailed aspects of this process		

3. <b>Types of OMICs data:</b> types of data needed in various research projects	<ul style="list-style-type: none"> <li>• Practical examples</li> <li>• Case-study discussions</li> </ul>	
4-5. <b>RNA-seq:</b> mapping techniques, counting (FPKM, RPKM), and statistical comparison		
6-7. <b>DNA-seq:</b> introduction to the concept of variant calling (SNPs) and annotation		
8-10. <b>Concept of Single Cell:</b> basics of single-cell level analysis		
11. <b>Microbiome</b> analysis: elements of metagenomics		
12-14. <b>Applications:</b> data retrieval from the Sequence Read Archive (SRA), quality control, mapping, statistics, variant calling, pathway analysis, and drug discovery		
<b>Bibliography</b>		
1. Course notes		
<b>8.2 Seminar / laboratory</b>	<b>Teaching methods</b>	<b>Remarks</b>
1. Introduction to the seminars/laboratories on Applied Genomics in Human Health. Syllabus and educational objectives.	<ul style="list-style-type: none"> <li>· Interactive exposure</li> <li>· Problem-solving activities</li> <li>· Hands-on case-study</li> <li>• Team work activities</li> </ul>	
2. Genome assembly: introduction to graphs and existing methods		
3. Transcriptome assembly: introduction to graphs and existing methods		
4-5. RNA-seq: mapping techniques, counting (FPKM, RPKM), and statistical comparison.		
6-7. DNA-seq: introduction to the concept of variant calling (SNPs) and annotation		
8-10. Single-cell concept: basics of single-cell level analysis		
11. Microbiome analysis: elements of metagenomics		
12-14. Applications: data retrieval from Sequence Read Archive (SRA), quality control, mapping, statistics, variant calling, metabolic pathway analysis, and drug discovery		
<b>Bibliography</b>		
1. Seminar notes		
2. <a href="https://www.sc-best-practices.org/preamble.html">https://www.sc-best-practices.org/preamble.html</a>		
3. <a href="https://www.biostarhandbook.com/index.html">https://www.biostarhandbook.com/index.html</a>		
4. Pevsner J. (2015) Bioinformatics and Functional Genomics, 3rd Ed. Blackwell Pub, UK.		

**9. Corroborating the content of the discipline with the expectations of the epistemic community, professional associations and representative employers within the field of the program**

<ul style="list-style-type: none"> <li>• The course has a similar content to courses from other European universities, being constantly updated and adapted to the level of training of students.</li> </ul>
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**10. Evaluation**

Type of activity	10.1 Evaluation criteria	10.2 Evaluation methods	10.3 Share in the grade (%)
10.4 Course	Knowledge of informational content	Written exam	60%
10.5 Seminar/lab activities	Ability to interpret cellular and molecular biology data in the field	Evaluation of annt	40%
10.6 Minimum performance standards			

Each student should be graded at least 5 (five) at both the course and the practical works assessments.

Date

Signature of course coordinator

Signature of seminar coordinator

**10.07.2024**

**Rareș Călin Lucaciu**

**Rareș Călin Lucaciu**

**Lect. Cruceriu Daniel, PhD**



**Lect. Cruceriu Daniel, PhD**



Date of approval

Signature of the head of department

**16.07.2024**

**Assoc. Prof. Beatrice Kelemen**