SYLLABUS

Applied genomics in human health

Academic year 2025-2026

1. Information regarding the programme

1.1. Higher education institution	Babeș-Bolyai University
1.2. Faculty	Faculty of Biology and Geology
1.3. Department	Department of Molecular Biology and Biotechnology
1.4. Field of study	Biology
1.5. Study cycle	Master, 4 semesters
1.6. Study programme/Qualification	Medical Biology Biologist
1.7. Form of education	Full-time

2. Information regarding the discipline

2.1. Name of the discipline	**Applied genomics in human health**					Discipline code	**BME 1132**
2.2. Course coordinator				Lecturer Cruceriu Daniel, PhD			
2.3. Seminar coordinator				Lecturer Cruceriu Daniel, PhD			
2.4. Year of study	2	2.5. Semester	3	2.6. Type of evaluation	E	2.7. Discipline regime	Optional

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

3.1. Hours per week	**4**	of which: 3.2 course	**4**	3.3 seminar/laboratory	**4**
3.4. Total hours in the curriculum	56	of which: 3.5 course	28	3.6 seminar/laborator	**28**
Time allotment for individual study (ID) and self-study activities (SA)					**hours**
Learning using manual, course support, bibliography, course notes (SA)					20
Additional documentation (in libraries, on electronic platforms, field documentation)					20
Preparation for seminars/labs, homework, papers, portfolios and essays					16
Tutorship					8
Evaluations					4
Other activities: two-way communication with the course holder / tutor					2
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	• Cell and molecular biology, Genetics, Biostatistics
4.2. competencies	• Interpretation of cell and molecular biology data • Beginner programming skills (bash and R)

5. Conditions (if necessary)

5.1. for the course	Online meeting platform Beamer, projection screen
5.2. for the seminar / lab activities	Attendance of a minimum of 90% of seminar classes is mandatory for granting the participation at the written exam. Computers, specific environment for developing and implementing bioinformatic pipelines/tools

6. Specific competencies acquired [1]

Professional competencies	• Applying OMICs data analysis methods (DNA-seq, RNA-seq, single-cell) to investigate the molecular mechanisms involved in human health and disease. • Developing and implementing bioinformatics pipelines for the analysis of genetic variations, differential gene expression, and the involved molecular pathways. • Using genomic and transcriptomic assembly algorithms, as well as specialized software, for the reconstruction and interpretation of sequencing data. • Analyzing the human microbiome through amplicon and metagenomic sequencing methods, evaluating its impact on health status.
Transversal competencies	• Application of efficient work rules and responsible attitudes towards the scientific domain, for the creative exploitation of one's own potential according to the principles and rules of professional ethics • Efficient conduct of activities organized in an interdisciplinary group and development of empathic capacity of interpersonal communication, networking and collaboration with diverse groups • Use of efficient methods and techniques for learning, information, research and development of abilities for knowledge exploitation, for adapting to the needs of a dynamic society and for communication in English.

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

7.1 General objective of the discipline	• Acquiring advanced theoretical knowledge and practical skills in the analysis and interpretation of OMICs genomic and transcriptomic data, with applicability in biomedical research and personalized medicine.
7.2 Specific objective of the discipline	• Familiarizing students with the technological and methodological principles of next-generation sequencing (NGS), as well as the types of OMICs data used in biomedical studies. • Developing skills in building and running bioinformatics workflows for genomic, transcriptomic, and metagenomic analysis, using specialized software. • Training the ability to biologically interpret results obtained from OMICs analyses, with a focus on identifying clinically relevant genetic variations, differential expression, and molecular regulatory networks involved in human diseases.

8. Content

8.1 Course	**Teaching methods**	**Remarks**
1. **Introduction** to the Applied Genomics in Human Health course: syllabus and educational objectives.	• Interactive exposure • Presentation • Explanation • Practical examples • Case-study discussions	
2. **Sequencing**: introduction to the concept, operation of a sequencing machine, and detailed aspects of this process		
3. **Types of OMICs data**: types of data needed in various research projects		
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. **Concept of Single Cell**: basics of single-cell level analysis		
11. **Microbiome** analysis: elements of metagenomics		

[1] One can choose either competences or learning outcomes, or both. If only one option is chosen, the row related to the other option will be deleted, and the kept one will be numbered 6.

12-14. Theoretical basics on data retrieval from the Sequence Read Archive (SRA), quality control, mapping, statistics, variant calling, pathway analysis, and drug discovery		

Bibliography

1. Ahmed, Z., Zeeshan, S., Mendhe, D., & Dong, X. (2020). Human gene and disease associations for clinical-genomics and precision medicine research. Clinical and translational medicine, 10(1), 297-318.
2. Carrasco-Ramiro, F., Peiró-Pastor, R., & Aguado, B. (2017). Human genomics projects and precision medicine. Gene therapy, 24(9), 551-561.
3. Roberts, M. C., Fohner, A. E., Landry, L., Olstad, D. L., Smit, A. K., Turbitt, E., & Allen, C. G. (2021). Advancing precision public health using human genomics: examples from the field and future research opportunities. Genome medicine, 13(1), 97.
4. Course notes

The above references are available online through the branch libraries of the Central University Library of Cluj-Napoca.

8.2 Seminar / laboratory	**Teaching methods**	**Remarks**
Introduction to the seminars/laboratories on Applied Genomics in Human Health. Syllabus and educational objectives.	Interactive exposure Problem-solving activities Hands-on case-study Teamwork activities	
Practical exercises on genomics and transcriptomics applied to human health.		
Practicals on data retrieval from Sequence Read Archive (SRA), quality control, mapping, statistics, variant calling, metabolic pathway analysis, and drug discovery		
Presentation of a project in the topic of the course	Evaluation	

Bibliography

1. Seminar notes
2. https://www.sc-best-practices.org/preamble.html
3. https://www.biostarhandbook.com/index.html
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

- The content of this course is relevant for conducting professional research activities in academic settings, as well as in public and private entities with research and development departments in the biomedical and human health fields.
- The course content is similar to courses offered at other European universities, being continuously updated and adapted to the students' level of preparation.. .

10. Evaluation

Type of activity	10.1 Evaluation criteria	10.2 Evaluation methods	10.3 Share in the grade (%)
10.4. Course	Knowledge of concepts and methods applied in human genomics	Written exam	50%
10.5. Seminar/lab activities	Apply methods for human genomics in real-life problems	Project implementation and presentation	50%
10.6. Minimum performance standards			
Each student should obtain a minimal grade of 5 (five) for the written exam and for the final grade. To obtain the minimum grade 5, the student must demonstrate the mastery of the basic concepts described during the course and seminars/practical classes, respectively.			

11. Labels ODD (Sustainable Development Goals)[2]

	SDG 3. Health and well-being

Date:
08.01.2025

Signature of course coordinator

Lecturer Cruceriu Daniel, PhD

Signature of seminar coordinator

Lecturer Cruceriu Daniel, PhD

Date of approval:
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Signature of the head of department

Assoc. Prof. Beatrice Kelemen, PhD

[2] Keep only the labels that, according to the *Procedure for applying ODD labels in the academic process*, suit the discipline and delete the others, including the general one for *Sustainable Development* – if not applicable. If no label describes the discipline, delete them all and write „*Not applicable*.".