

SYLLABUS

1. Information regarding the programme

1.1 Higher education institution	Babeş 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
1.6 Study programme / Qualification	Bioinformatics applied in life sciences

2. Information regarding the discipline

2.1 Name of the discipline (en) (ro)	Applied Genomics in human health Genomică aplicată în sănătatea umană						
2.2 Course coordinator	Sef lucr. Dr. Cruceriu Daniel Dr. Armean Irina						
2.3 Seminar coordinator	Sef lucr. Dr. Cruceriu Daniel Dr. Armean Irina						
2.4. Year of study	2	2.5 Semester	2	2.6. Type of evaluation	C	2.7 Type of discipline	Elective
2.8 Code of the discipline	BME1132						

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

3.1 Hours per week	4	Of which: 3.2 course	2	3.3 seminar/laboratory	2
3.4 Total hours in the curriculum	56	Of which: 3.5 course	28	3.6 seminar/laboratory	28
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"> • Cell and molecular biology • Genetics, genomics and functional genomics • Biostatistics • Fundamentals of programming
4.2. competencies	<ul style="list-style-type: none"> • Interpretation of cell and molecular biology data • Beginner programming skills (bash and R)

5. Conditions (if necessary)

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

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

Professional competencies	<ul style="list-style-type: none"> • To interpret the raw data obtained from the analyzes and techniques used in molecular diagnosis in oncology. • To interpret the raw data obtained from the analyzes and techniques used in research in the field of oncobiology, oncogenetics, oncogenomics. • To interpret data processed from the specialized literature in the field of oncobiology, oncogenetics, oncogenomics. • To use biological databases dedicated to various human pathologies.
Transversal competencies	<ul style="list-style-type: none"> • To use the theoretical notions in solving practical problems in molecular diagnosis and research in the field of oncobiology, oncogenetics, oncogenomics and other human pathologies • To carry out the transfer of information for the understanding of the human genome, in general, and oncogenomics, in particular, by taking over and using knowledge from related fields: cytology, genetics, molecular biology, biostatistics and bioinformatics.

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

7.1 General objective of the discipline	<ul style="list-style-type: none"> • Knowledge of the main technologies of human genome sequencing, biological databases dedicated to human pathologies and the main approaches in the field of oncobiology, oncogenetics and oncogenomics both in the clinic (laboratory for the molecular diagnosis of cancer) and in research.
7.2 Specific objective of the discipline	<ul style="list-style-type: none"> • To comparatively explain (normal cell-tumor cell) the 6 characteristics of cancer from a cellular and molecular perspective. • To explain the principles of genetic and genomic testing (diagnosis and prognosis) in cancer. • To explain the principles of personalized medicine and targeted cancer therapies. • To interpret experimental and clinical data obtained by molecular biology methods specific to oncobiology, oncogenetics and oncogenomics. • To understand the principles of mass sequencing technologies applied to the human genome; • To use biological databases dedicated to various human pathologies.

8. Content

8.1 Course	Teaching methods	Remarks
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1. Introduction to "Applied Genomics in Human Health": syllabus and educational objectives; Cancer: definitions, epidemiology and current status.	<ul style="list-style-type: none"> • Interactive exposure • Presentation • Explanation • Practical examples • Case-study discussions 	
2. Introduction to oncogenomics: the central dogma of cell biology; introduction to cellular signalling.		
3. Characteristics of cancer: 1. Unlimited replication potential; 2. Loss of the ability to regulate the cell cycle (tumour suppressor genes in cancer) 3. Induction of proliferation (oncogenes in cancer).		
4. Characteristics of cancer: 4. Resistance to apoptosis. 5. Promoting of angiogenesis; 6. Invasive and metastatic capacity.		
5. Principles of genetic testing of cancer in current clinical practice (diagnosis and prognosis).		
6. Targeted cancer therapy and personalized medicine in current clinical practice: principles; small molecule drugs and monoclonal antibodies.		
7. Targeted cancer therapy and personalized medicine in current clinical practice: examples from the clinic (hormone therapies; tyrosine kinase inhibitors; angiogenic inhibitors; immunotherapy).		
8. Genomic cancer testing: principles		
9. Identifying mutations in cancer: sequencing I.		
10. Identifying mutations in cancer: sequencing II.		
11. Identifying epigenetic changes in cancer: microarray technique I.		
12. Identifying epigenetic changes in cancer: microarray technique II.		
13. DNA sequencing and large-scale sequencing of the human genome. Databases for human DNA variability		
14. Case studies (LRRK2 and Parkinson's) and databases used (gnomAD, Ensembl Variation, ClinVar).		
Bibliography		
1. Course notes		
2. Weinberg RA, 2013. The Biology of Cancer Second Edition, Garland Science, New York, USA		
3. Pecorino, L, 2005. Molecular Biology of Cancer, Oxford University Press, New York, USA		
4. Hanahan D, Weinberg RA, 2000. The hallmarks of cancer. Cell, 100(1):57-70		
5. Hanahan D, Weinberg RA, 2011. Hallmarks of cancer: the next generation. Cell, 144(5):646-674		
6. Kreso A, Dick JE, 2014. Evolution of the cancer stem cell model. Cell Stem Cell, 14(3):275-291.		
8.2 Seminar / laboratory	Teaching methods	Remarks
1. Introduction to the seminars/laboratories on Applied Genomics in Human Health. Syllabus and educational objectives.	<ul style="list-style-type: none"> • Interactive exposure • Problem-solving activities • Hands-on case-study • Team work activities 	
2. The Cancer Genome Atlas Database (TCGA): operating principles and working interface.		
3. The Cancer Genome Atlas Database (TCGA): applications.		
4. Polymerase chain reaction (PCR): identification of RAS gene mutations in colorectal cancer. RT-PCR: identification of BCR-ABL fusions in leukemias (interpretation of raw data in clinical diagnosis)		
5. qPCR: identification of EGFR gene mutations in lung cancer (interpretation of raw data in clinical diagnosis)		
6. RT-qPCR: determination of gene expression levels in cancer. (interpretation of raw data)		

7. Sequencing I: Characterization of the molecular signature of blood exosomes in patients with metastatic breast cancer (interpretation of raw data)		
8. Sequencing II: Characterization of the molecular signature of blood exosomes in patients with metastatic breast cancer (interpretation of raw data)		
9. Microarray I technique: exploring the mechanisms of intrinsic resistance to radiochemotherapy in cervical cancer (interpretation of raw data (GeneSpring / R))		
10. Microarray II technique: exploring the mechanisms of intrinsic resistance to radiochemotherapy in cervical cancer (interpretation of raw data (GeneSpring / R))		
11-12. Analysis of data from the literature. Oral presentations.		
13-14. Exercises for analyzing the data of human genetic variants from the sequencing of human genomes / exomes using Ensemble Variant Effect Predictor		
Bibliography		
1. McLaren, W., Gil, L., Hunt, S. E., Riat, H. S., Ritchie, G. R., Thormann, A., ... & Cunningham, F. (2016). The ensembl variant effect predictor. <i>Genome biology</i> , 17(1), 1-14. https://doi.org/10.1186/s13059-016-0974-4		

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 course has a similar content to courses from other European universities, being constantly updated and adapted to the level of training of students.
- The course aims to train the skills in the field of human genomics with focus on oncobiology, oncogenetics and oncogenomics in accordance with the European syllabus for the training of specialists in the medical laboratory (EC4 European Syllabus for Post-Graduate Training in Clinical Chemistry and Laboratory Medicine).
- The course contents are constructed taking into account the responsibilities of the biologist / biochemist in a medical laboratory for molecular diagnosis of cancer and research units in oncobiology / oncogenetics / oncogenomics, in accordance with the job description.

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 colloquium	60%
	Ability to use information in a new context		
	Ability to interpret molecular diagnostic analyzes and experimental raw / processed data		
10.5 Seminar/lab activities	Ability to interpret cellular and molecular biology data in the field	Oral presentation	15%
	Individual performance during laboratory/ seminar activities	Evaluation of the results obtained in workshops and homework	25%
10.6 Minimum performance standards			
Knowledge of 50% of the material contained in the course.			

Knowledge of 50% of the material from the practical works.

Date	Signature of course coordinator	Signature of seminar coordinator
16.01.2023	Lect. Dr. Cruceriu Daniel Dr. Armean Irina	Lect. Dr. Cruceriu Daniel Dr. Armean Irina

Date of approval	Signature of the head of department
20.01.2023	Assoc. Prof. Beatrice Kelemen