

1.DISCIPLINE SHEET

1. Program data

1.1 Higher education institution	UNIVERSITY OF MEDICINE AND PHARMACY "VICTOR BABEȘ" TIMIȘOARA
1.2 Faculty	FACULTY OF MEDICINE
1.3 Department	II
1.4 Field of study of Bachelor ¹⁾	Medicine
1.5 Study cycle ²⁾	License
1.6 Study program/ Qualification	Medicine

2. Discipline data

2.1. Name of the discipline	GENOMICS. CLINICAL BIOINFORMATICS							
2.2 Owner of course activities	Prof. Univ. Dr. Chiriță-Emandi Adela							
2.3 Holder of laboratory activities	Prof. Univ. Dr. Chiriță-Emandi Adela							
2.4 Year of study	II	2.5 The semester	II	2.6 Type of assessment	Colloquy	2.7 Discipline regime	Content ³⁾	DS
							Obligation ³⁾	DO

3. Time estimated total (hours per semester of teaching activities)

3.1 Number of hours per week	4	3.2 of which: course	2	3.3 practical application	2
3.4 Total hours from the curriculum	56	3.5 of which: course	28	3.6 practical application	28
The distribution of the time fund					hours
Study according to the textbook, course support, bibliography and notes					22
Additional documentation in the library, on specialized electronic platforms and in the field					10
Preparation of seminars / laboratories / projects, assignments, reports, portfolios and essays					10
Tutorial					
Exams (1 final exam)					1
Other activities					
3.7 Total hours of individual study	42				
3.8 Total hours per semester	100				
3.9 Number of credits ⁵⁾	1				

4. Prerequisites (where applicable)

4.1 of the curriculum	Medical genetics
4.2 skills	Clinical interpretation of a genomic analysis result

5. Terms (where applicable)

5.1 course development	<ul style="list-style-type: none"> Attendance at the course is highly desirable, a minimum of 7 attendances being required.
5.2 conducting the seminar/laboratory/practical application	<ul style="list-style-type: none"> Attendance at internships/practical work is mandatory, a maximum of 15% of the total absences being accepted. Recovery of paid absences during the last week of the semester is allowed within the limit of 15% of the total number of absences (with the exception of medical cases that will require individual Dean's approval).

6. Specific skills accumulated

Professional competences	<ol style="list-style-type: none"> Acquiring the terminology specific to Genomics Ability to describe principles of sequencing techniques Evaluation of a genomic variants Conception of counseling for a patient with a monogenic disease, following a genomic analysis result Knowing the limits of tests in genetic diseases
Transversal skills	<ol style="list-style-type: none"> Concern for professional improvement by training critical thinking skills demonstrated by active participation in the course and laboratory/seminar/project ; Involvement in scientific research activities by participating in the elaboration of reports, studies, specialist articles and participation in a research group Effective use of information sources and resources for communication and assisted professional training (Internet portals, specialized software applications, databases, online courses, etc.) both in Romanian and in an international language (English) ;

7. The objectives of the discipline (resulting from the specific skills acquired)

7.1 The general objective of the discipline	Acquisition of the fundamental notions of Genomics and clinical bioinformatics.
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	Genetic diseases are very different, rare, complex, this course offers information about the multidisciplinary approach model, teamwork, the use of databases and the approach to these diseases in the national and/or European network.
7.2 Specific objectives	Knowledge and understanding of genetic testing methods to obtain an accurate diagnosis. Differentiating monogenic from polygenic multifactorial diseases. the technical aspects of genetics and genomics research as well as their practical applications. Some of the topics include analysis of the human genome in psychiatric diseases and cancer, population genomics and pharmacogenomics.

8. Content

8.1 Course	Teaching methods	Number of hours	Remarks
1. Introduction to the human genome. Genetic diseases versus multifactorial diseases. Predictive medicine.	INTER-ACTIVE LECTURE	2	<ul style="list-style-type: none">● Oral lecture delivered with the help of interactive Powerpoint presentations, accompanied by rich and suggestive iconography, available on the university's Moodle e-learning platform.● The courses are presented in Power Point format, being systematized and accompanied by extremely rich iconography.● The course is structured according to the European teaching style and is updated annually with the latest information from international specialized literature.
2. Transcription. Translation. The architecture of a gene.		2	
3. Variations of the human genome (CNV, SNV). The reference genome. Classification of genomic variants (ISCN)		2	
4. Nomenclature of variants (HGVS). Classification of genomic variants (ACMG – introduction)		2	
5. Classification of genomic variants (ACMG-advanced, ABC System)		2	
6. Analysis of genomic data using bioinformatics techniques.		2	
7. Genomics in cancers - general aspects, germline testing		2	
8. Genomics in cancers - somatic testing		2	
9. Epigenetics/ epigenomics. GWAS studies		2	
10. Gene therapy		2	
11. Beyond genomics – other OMICS (transcriptomics, proteomics, metabolomics, epigenomics, phenomics, exposomics)		2	
12. Ethics in genomic testing		2	
13. Preimplantation genomic testing. Genomic neonatal screening		2	
14. Recapitulation		2	
Mandatory bibliography: 1. Medical Genetics course. Maria Puiu, Dorina Stoicănescu, Cristina Gug, Simona Farcas, Cristina Popa, Nicoleta Andreescu, Adela Chirita-Emandi, Andreea Dobrescu., 978-606-32-0296-4, Eurostampa Publishing House, Timisoara, 2016. 2. Medical genetics. Mircea Covic, Dragos Stefanescu, Ionel Sandovici, Vlad Gorduza. Polirom Publishing House, 2017 Optional bibliography: 1. New Clinical Genetics 3. Andrew Read, Dian Donnai. Scion Publ. Ltd, 2015 2. Harrison'S Principles of Internal Medicine 19 th edition Kasper D, Fauci A, Hauser S, Longo D, Jameson J. L, Loscalzo J , McGraw-Hill Education, 2015			
8.2 Laboratory/ practical application	Teaching-learning methods	Number of hours	Remarks
1. How do we choose a genetic test? The "genetic test" is a myth. Benefits and limitations of gene panel, exome or genome testing in monogenic diseases	LECTURE + DEBATE + CASE/ STUDY PRESENTATION / JOURNAL CLUB	2	<ul style="list-style-type: none">● Oral lecture delivered with the help of Powerpoint presentations available on the university's Moodle e-learning platform.● Presentation of cases. Diagnostic algorithms to guide the thinking of the future doctor directed towards the investigations necessary to establish a correct diagnosis.● Presentation of investigation methods for clinical, differential, etiological
2. Molecular genetic techniques (DNA extraction, PCR, MLPA, Array)		2	
3. Array variant classification exercises – variations in the number of CNV copies		2	
4. Molecular genetic techniques (sequencing)		2	
5. Genomic variants classification exercises - single nucleotide variants, SNVs		2	
6. Phenotypic significance of some variants and advice for clarifying the consequences of genomic variant analysis. How to understand a variant of uncertain significance in the clinic		2	

7. Genetic testing in cancers, germline testing – examples		2	diagnosis. Presentation of ethical principles, support groups for patients. ● Checking the mastery of the main knowledge taught through grid questions at the end of the PA.
8. Genetic testing in cancers, somatic testing – examples		2	
9. Examples of results for personalized medicine (nutrigenomics, pharmacogenomics, metagenomics)		2	
10. Journal Club 1: critical analysis of a genomics-related publication, Students presentation (20 minutes) and discussion of the paper presented		2	
11. Journal Club 2: critical analysis of a publication related to GWAS, OMICS Students presentation (20 minutes) and discussion of the presented paper		2	
12. Ethical dilemmas of species in genomics (group activity)		2	
13. Fertilization in vitro. Peculiarities in genomic testing in preimplantation diagnosis and genomic neonatal screening		2	
14. Recapitulation		2	
Mandatory bibliography: Genetics - Practical Applications. Maria Puiu, Dorina Stoicanescu, Cristina Gug, Simona Farcas, Cristina Popa, Nicoleta Andreescu, Adela Chirita-Emandi, Andreea Dobrescu, Alexandra Mihailescu. Eurostamp Publishing House, 2019.			
Optional bibliography: 1. Smith's Recognizable Patterns of Human Malformation, K. Jones. Saunders, 2013			

Mandatory bibliography:

Optional bibliography:



9. Corroboration of the contents of the discipline with the expectations of representatives of the epistemic communities, professional associations and representative employers in the field related to the program

Genomic knowledge will allow future doctors to identify the genetic causes of diseases, regardless of the specialty they will practice. Knowing the aspects of predictive diagnosis, ethical, legal and social issues in genetic diseases will allow them to have an interdisciplinary approach to the genetic pathology of the genomic cause.

Information on pharmacogenomics, nutrigenomics and oncogenomics (personalized medicine) will enable today's students to become highly competent physicians. They will be able to provide collaborating physicians with information about the effect of drugs based on patients' genetic testing. Personalized medicine is the medicine of the future. Knowledge of genomics will be an asset in medical practice and career development.

10. Evaluation

Type of activity	10.1 Evaluation criteria	10.2 Evaluation methods	10.3 Weight of the final grade
10.4 Course	<p><i>Knowledge for grade 5:</i> The student must demonstrate knowledge of the principles of genomic testing methods and their applicability</p> <p><i>Knowledge for grade 10:</i> The student must have a thorough knowledge of genomic testing methods, test limits, methods of interpreting genomic variants, general concepts of epigenetics and OMICS.</p>	<i>Final assessment :</i> MCQ test with 50 questions, during 1 hour	50%
10.5 Practical applications	<p><i>Knowledge for grade 5:</i> The student must demonstrate an understanding of the principles of the significance of genomic variants in the clinic.</p> <p><i>Knowledge for grade 10:</i> The student must recognize the limits of genomic tests, understand the use of the main tools for the analysis of genomic variants, know the principles of the significance of genomic variants in the clinic, know aspects related to bioinformatics and ethical concepts in genomic testing.</p>	<p><i>Continuous assessment:</i> appreciation of student activities during the semester, focusing on aspects of knowledge, synthesis and application of information</p> <p><i>Final assessment:</i> practical exam</p>	<p>10%</p> <p>40%</p>
10.6 Minimum Performance Standard			
Familiarization with notions of genomics.			

Date of completion 15.04. 2024	Course coordinator's signature Prof. Univ. Dr. Chiriță-Emandi Adela 	Signature of the practical application coordinator Prof. Univ. Dr. Chiriță-Emandi Adela 
The signature of the head of discipline Prof. Dr. Puiu Maria		
Date of approval in the department 20.04. 2024	Signature of the department director Prof. Dr. Dema Alice	

Note :

- ¹⁾ Field of study - *choose one of the options:* Bachelor's/Master's/PhD (**to be completed according to the Nomenclature of fields and specializations/university study programs in force**);
- ²⁾ Study cycle - *choose one of the options:* Bachelor's/ Master's/ Doctorate;
- ³⁾ Discipline regime (content) - *choose one of the options:* **DF** (fundamental discipline)/ **DD** (discipline in the field)/ **DS** (specialized discipline)/ **DC** (complementary discipline) - *for the undergraduate level*; **DAP** (deepening discipline)/ **DSI** (synthesis discipline)/ **DCA** (advanced knowledge discipline) - *for the master's level*;
- ⁴⁾ Discipline regime (compulsory) - *choose one of the options:* **DI** (compulsory discipline)/ **DO** (optional discipline)/ **DFac** (facultative discipline);
- ⁵⁾ One credit is equivalent to 25-30 study hours (teaching activities and individual study) .
- ⁶⁾ For the specializations and/or subjects whose subject is found in the residency bibliography, this becomes mandatory.