

FILE OF DISCIPLINE / COURSE SYLLABUS

1. Information regarding the Study Program

1.1 Higher Education Institution	"VICTOR BABEȘ" UNIVERSITY OF MEDICINE AND PHARMACY OF TIMIȘOARA
1.2 Faculty	MEDICINE
1.3 Department	II
1.4 Study Domain ¹⁾	MEDICINE
1.5 Study Cycle ²⁾	Bachelor
1.6 Study program/ Qualification	Medicine

2. Information regarding the Discipline

2.1.Name of the discipline	GENOMICS. CLINICAL BIOINFORMATICS							
2.2 Course coordinator	Prof. Dr. Chiriță-Emandi Adela							
2.3 Practical activity/laboratory coordinators	Prof. Dr. Chiriță-Emandi Adela							
2.4. Year of study	II	2.5 Semester	4	2.6 Type of assessment	Colloquy	2.7 Course rank/Type of discipline	Content ³⁾	DS
							Mandatory /Compulsory ³⁾	DO

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

3.1 Number of hours/ week	4	Of which: 3.2 lecture/course	2	3.3 Practical activity/laboratory	2
3.4 Total hours in the curriculum	56	Of which: 3.5 lecture/course	28	3.6 Practical activity/laboratory	28
Time allotment:					hours
Learning using manuals, lecture support, bibliography and lecture notes					10
Additional documentation – in the library, dedicated electronic platforms, field documentation					4
Preparation for seminars/ practical activity/ projects, homework, papers, portfolios and essays					4
Tutoring					
Evaluations					1
Other activities					
3.7 Total individual study hours	18				
3.8 Total hours per semester	hours				
	75				
3.9 Number of ECTS credits ⁵⁾	3				

4. Prerequisites (if necessary)

4.1 Courses - studied curriculum	Genetics
4.2 Competencies/skills/abilities	

5. Conditions (if necessary)

5.1 For the courses/lectures	<ul style="list-style-type: none"> Attendance at the course is mandatory, a maximum of 50% of the total absences being accepted. Oral lectures delivered with the help of interactive Powerpoint presentations, accompanied by rich and suggestive iconography, case presentations.
5.2 For the laboratory/practical activity/project	<ul style="list-style-type: none"> Attendance at internships/practical work is mandatory, a maximum of 70% of total absences being accepted. Recovery is allowed within the limit of 30% of the total number of paid absences in the last week (except for medical cases that will require individual Dean's approval). Interactive presentations, case presentations. Diagnostic algorithms to guide the thinking of the future doctor directed towards the investigations necessary to establish a correct diagnosis.

6. Specific competencies acquired

Professional Competencies	<ol style="list-style-type: none"> Acquiring the terminology specific to Genomics. Ability to describe principles of sequencing techniques. Evaluation of 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.
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Transversal Competencies	1. Concern for professional improvement by training critical thinking skills demonstrated by active participation in the course and laboratory/seminar/project.
	2. Involvement in scientific research activities by participating in the elaboration of reports, studies, specialist articles and participation in a research group.
	3. 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. Objectives of the discipline (outcome of the acquired competencies)

7.1 Discipline/Course general objectives	1. Acquisition of the fundamental notions of Genomics and clinical bioinformatics. 2. Genetic diseases are very different, rare, and 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 Discipline/Course specific objectives	1. Knowledge and understanding of genetic testing methods to obtain an accurate diagnosis. 2. 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.	INTERACTIVE LECTURE	2	<ul style="list-style-type: none"> 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. Digital tools used in clinical genomics		2	

Mandatory bibliography:⁶⁾

Lecture notes in ppt/pdf format. The information is available on the Moodle e-learning platform on the university website.

Optional bibliography:

1. New Clinical Genetics 3. Andrew Read, Dian Donnai. Scion Publ. Ltd, 2015
2. Harrison'S Principles of Internal Medicine 19th edition Kasper D, Fauci A, Hauser S, Longo D, Jameson J. L, Loscalzo J, McGraw-Hill Education, 2015

8.2 Seminar/ Laboratory/practical activity/ projects	Teaching and 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	LECTURE +	2	<ul style="list-style-type: none"> Oral lecture delivered with the help of Powerpoint

testing in monogenic diseases	DEBATE + CASE/ STUDY PRESENTATION/ JOURNAL CLUB		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 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.
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	
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. In vitro fertilization. Peculiarities in genomic testing in preimplantation diagnosis and genomic neonatal screening		2	
14. Recapitulation		2	

Mandatory bibliography:
Notes in ppt/pdf format. The information is available on the Moodle e-learning platform on the university website.

Optional bibliography:
1. Smith's Recognizable Patterns of Human Malformation. K. Jones. Saunders, 2013

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

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.

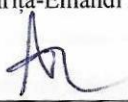
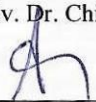
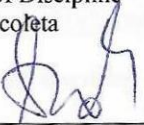

Information on pharmacogenomics, nutrigenomics and oncogenomics (personalized medicine) will enable today's students to become highly competent physicians. They will be able to provide collaborating physicians with information about the effect of drugs based on patients' genetic testing. Personalized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

In order to outline and standardize the content, as well as the choice of teaching/learning methods, the holders of the discipline organized and participated in national and international Medical Genetics Conferences. The meetings were aimed at identifying the needs and expectations of employers in the field and coordinating with other similar programs at other medical universities. The information and skills acquired will allow him to face the current demands of the labor market in

the health field, meeting European educational and professional standards.

10. Assessment/Evaluation

Type of activity	10.1 Assessment criteria	10.2 Assessment methods	10.3 Percentage of 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 activity/ seminar	<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 standards-basic knowledge			
Familiarization with notions of genomics.			

Date 28.04.2025	Signature of the course coordinator Prof. Dr. Chiriță-Emandi Adela 	Signature of the practical application coordinator Prof. Univ. Dr. Chiriță-Emandi Adela 
Signature of the Head of Discipline Prof. Dr. Andreescu Nicoleta 		
Date of approval in the Department	Signature of the Head of Department Prof. Dr. Dema Alis 	

- 1) Domeniul de studii - se alege una din variantele: Licență/ Masterat/ Doctorat (se completează conform cu Nomenclatorul domeniilor și al specializărilor/ programelor de studii universitare în vigoare) ;
- 2) Ciclul de studii - se alege una din variantele: Licență/ Master/ Doctorat;

- 3) Regimul disciplinei (conținut) - *se alege una din variantele:* **DF** (disciplină fundamentală)/ **DD** (disciplină din domeniu)/ **DS** (disciplină de specialitate)/ **DC** (disciplină complementară) - *pentru nivelul de licență*; **DAP** (disciplină de aprofundare)/ **DSI** (disciplină de sinteză)/ **DCA** (disciplină de cunoaștere avansată) - *pentru nivelul de masterat*;
- 4) Regimul disciplinei (obligativitate) - *se alege una din variantele:* **DI** (disciplină obligatorie)/ **DO** (disciplină opțională)/ **DFac** (disciplină facultativă);
- 5) Un credit este echivalent cu 25 de ore de studiu (activități didactice și studiu individual).

*nr de ore de studiu individual (punctul 3.7.) = nr total ore (nr credite X 25) minus nr. ore din planul de învățământ (punctul 3.4) minus ore alocate pentru examinări. Aceste ore se împart între

Studiul după manual, suport de curs, bibliografie și notițe	
Documentare suplimentară în bibliotecă, pe platformele electronice de specialitate și pe teren	
Pregătire seminarii/ laboratoare/ proiecte, teme, referate, portofolii și eseuri	
Tutoriat	

- 6) Pentru specializările și/sau disciplinele a căror tematică se regăsește în bibliografia de rezidențiat, aceasta devine obligatorie. Dintre titlurile bibliografice, 50% trebuie să fie din ultimii 5 ani.

