

# 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 .....	MEDICINE
1.5 Study Cycle <sup>2)</sup>	Bachelor
1.6 Study program/ Qualification	Medicine

## 2. Information regarding the Discipline

2.1 Name of the discipline	Clinical Genetics							
2.2 Course coordinator	Conf. Dr. Stoicănescu Dorina							
2.3 Practical activity/laboratory coordinators	Conf. Dr. Stoicănescu Dorina							
2.4 Year of study	VI	2.5 Semester	1	2.6 Type of assessment	Colloquy	2.7 Course rank/Type of discipline	Content <sup>3)</sup>	SD
			1				Mandatory /Compulsory <sup>3)</sup>	FacD

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

3. Total estimated time (number of hours/semester or equivalent activities)					
3.1 Number of hours/week	2	Of which: 3.2 lecture/course	1	3.3 Practical activity/laboratory	1
3.4 Total hours in the curriculum	28	Of which: 3.5 lecture/course	14	3.6 Practical activity/laboratory	14
Time allotment:					hours
Learning using manuals, lecture support, bibliography and lecture notes					
Additional documentation – in the library, dedicated electronic platforms, field documentation					
Preparation for seminars/ practical activity/ projects, homework, papers, portfolios and essays					
Tutoring					
Evaluations (colloquy)					1
Other activities					
3.7 Total individual study hours	1				
3.8 Total hours per semester	29 hours				
3.9 Number of ECTS credits <sup>5)</sup>	1				

## 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"> <li>Attendance at the course is mandatory, a maximum of 50% of the total absences being accepted.</li> <li>Oral lectures delivered with the help of interactive Powerpoint presentations, accompanied by rich and suggestive iconography, case presentations.</li> </ul>
5.2 For the laboratory/practical activity/project	<ul style="list-style-type: none"> <li>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).</li> <li>Interactive presentations, case presentations. Diagnostic algorithms to guide the thinking of the future doctor directed towards the investigations necessary to establish a correct diagnosis.</li> </ul>

## 6. Specific competencies acquired

Professional Competencies	<ol style="list-style-type: none"> <li>Acquiring the terminology specific to Clinical Genetics.</li> <li>The ability to describe the clinical picture of the main genetic syndromes as well as the correlation with genetic tests.</li> <li>Evaluation of patients with genetic diseases or disorders with a genetic component.</li> <li>Designing a test algorithm in genetic diseases.</li> <li>Knowledge of the teratogenic effects of drugs; specification of allowed/forbidden drugs during pregnancy.</li> </ol>
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<b>Transversal Competencies</b>	<ol style="list-style-type: none"> <li>1. Concern for professional improvement by training critical thinking skills demonstrated by active participation in the course and laboratory/seminar/project.</li> <li>2. Involvement in scientific research activities by participating in the elaboration of reports, studies, specialized articles and participation in a research group.</li> <li>3. The 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.</li> </ol>
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#### 7. Objectives of the discipline (outcome of the acquired competencies)

7.1 Discipline/Course general objectives	Students will acquire the theoretical and conceptual tools used in Clinical Genetics. Students will learn the multidisciplinary approach model, teamwork, the use of databases and the approach to genetic diseases in the national and/or European network.
7.2 Discipline/Course specific objectives	<p>Knowing and understanding the signs and symptoms of genetic pathology, structured by life cycles and correlated with genetic testing methods to obtain an accurate diagnosis.</p> <p>Knowledge and understanding of case management methods from clinical suspicion to disease diagnosis, with the possibility of antenatal diagnosis, treatment methods, recovery and prevention of genetic diseases, calculation of recurrence risk in monogenic diseases</p> <p>Differentiating monogenic from polygenic multifactorial diseases.</p>

#### 8. Content

8.1 Course	Teaching methods	Number of hours	Remarks
1. Fundamentals of molecular and cellular human mechanisms of heredity. Types of genetic tests that are used to diagnose genetic diseases.	INTERACTIVE LECTURE	2	<ul style="list-style-type: none"> <li>• Available on University's Moodle e-learning platform</li> <li>• The course is structured according to the European manner of teaching, is updated annually with the latest information from international specialized literature.</li> </ul>
2. Genetic pathology and prenatal period. Genetic Testing in Prenatal Diagnosis. Aspects of normal fetal development and the role of teratogenic factors.		2	
3. Genetic conditions relevant to newborns and children. Clinical aspects in dysmorphology. Diagnosis of major congenital genetic diseases and/or with onset during infancy or childhood.		2	
4. Genetic pathology in teenagers and adults. Genetic mechanisms of predisposition in common diseases. Genetic diseases or having genetic components with late onset. Aspects of predictive diagnosis.		2	
5. Interdisciplinary approach to genetic pathology (oncologic, cardiovascular, dermatologic pathology).			
6. Interdisciplinary approach to genetic pathology (ENT, ophthalmic, endocrine, gynecologic, neuropsychiatric pathology, etc.)		2	
7. Ethical, social, and legal issues in Clinical Genetics. Genetic testing. The role and place of research in genetic pathology. Methods of communication with the patient, family, collaborators and specialized laboratories.		2	

##### **Mandatory bibliography:** <sup>6)</sup>

1. New Clinical Genetics 4<sup>th</sup> ed. Andrew Read, Dian Donnai. Scion Publ. Ltd, 2020

##### **Optional Bibliography:**

1. Harrison's Principles of Internal Medicine 20th edition Kasper D, Fauci A, Hauser S, Longo D, Jameson J. L, Loscalzo J, McGraw Hill / Medical; 2018

8.2 Seminar/ Laboratory/practical activity/ projects	Teaching and learning methods	Number of hours	Remarks
1. Characteristics of family history	LECTURE	2	



in genetic counseling. Identification of different inheritance patterns in the family trees. Establishment of genetic risks. Genetic technologies for diagnosis and research.	<p style="text-align: center;">+ DISCUSSION + STUDIES - CASE STUDIES</p>		<ul style="list-style-type: none"> <li>• Case presentations. Diagnostic algorithms to guide thinking of the future medical practitioner toward targeted investigations necessary to establish a correct diagnosis.</li> </ul>
2. Characteristics of family history in prenatal diagnosis. Indications and methods of prenatal diagnosis. Organization of prenatal genetic services. Ethical, legal and social issues in prenatal diagnosis.		2	<ul style="list-style-type: none"> <li>• Presentation of investigative methods for clinical, differential etiological diagnosis. Ethical principles, support groups for patients.</li> </ul>
3. Somatometric indices tracked in dysmorphisms, ways of recording them. How to use journals and databases in dysmorphology. Characteristics of family history in dysmorphology. Screening programs during the neonatal period.		2	<ul style="list-style-type: none"> <li>• Verifying the acquisition of the main knowledge by multiple choice questions at the end of the lab.</li> </ul>
4. Aspects of genetic epidemiology and biostatistics. Role of patient education and of family physician in the management of common diseases. Ethical, legal and social issues in common diseases of the adult. Methods of genetic counseling.		2	
5. Current recommendations on the supervision of family cancer-prone individuals. Methods of genetic testing in the main types of cancer.		2	
6. Main clinical aspects, genetic investigation, management of some multifactorial diseases. How to work with other specialists, as a member in a multidisciplinary team.		2	
7. European legislation related to rare diseases. The importance of support groups for patients and family. Ethical principles governing genetic testing, informed consent procedures. Communication with patients, family, collaborators and specialized laboratories. Legal opportunities and social assistance for people with disabilities.		2	

**Mandatory bibliography:**

1. Synopsis of Medicine, Latha Ganti, David Lebowitz, Javier Rosario, Ariel Vera, Romanian edition coordinators: Cristina Oana Mărginean, Cătălina Poiană, 5th Edition, Hipocrate Publishing House, Bucharest, 2021
2. Kumar & Clark Clinical Medicine, Kumar, Clark, 10th Ed. Elsevier, 2020
3. Thompson & Thompson Genetics in Medicine, Nussbaum, McInnes, Willard 9th Ed. Elsevier, 2019
4. Harper's Practical Genetic Counselling, Eighth Edition. A. Clarke, CRC Press, 2019

**Optional Bibliography:**

1. Smith's Recognizable Patterns of Human Malformation, K. Jones. Saunders, 2021





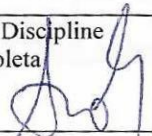
**9. Corroborating the content of the discipline with the expectations of the epistemic community, professional associations and representative employers within the field of the program**

Knowledge of clinical genetics 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 genetic pathology. Knowledge of clinical genetics will be an advantage 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>For grade 5</i> Students must prove knowledge of the main signs in genetic and multifactorial disorders, inheritance patterns, and interpreting a family tree</p> <p><i>For grade 10</i> Students must have a thorough knowledge of genetic and partially genetic disorders, clinical features, positive and differential diagnosis, treatment and prevention options, genetic risk assessment, genetic testing methods.</p>	<p><i>Continuous evaluation</i></p> <p><i>Final evaluation:</i> Colloquy: editorial topic</p>	50%
10.5 Practical activity/ seminar	<p><i>For grade 5</i> students must prove knowledge of main disorders with a genetic component, technologies for diagnosis, prenatal diagnosis, ethical principles governing genetic testing.</p> <p><i>For grade 10</i> students should recognize certain pathological phenotypes, draw and interpret family trees, prepare a plan for managing a patient, aspects of genetic epidemiology, biostatistics, legislation related to rare diseases.</p>	<p><i>Continuous evaluation</i></p> <p><i>Final evaluation:</i> editorial topic</p>	50%
10.6 Minimum performance standards-basic knowledge			
Familiarizing with clinical genetics notions. Knowledge and understanding of genetic terminology.			

Date 28.04.2025	Signature of the course coordinator Conf. Dr. Stoicănescu Dorina 	Signature of the laboratory/seminar coordinator Conf. Dr. Stoicănescu Dorina 
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.



