

FIȘA DISCIPLINEI

1. Date despre program

1.1 Instituția de învățământ superior	UNIVERSITY OF MEDICINE AND PHARMACY "VICTOR BABEȘ" TIMIȘOARA
1.2 Facultatea	FACULTY OF MEDICINE
1.3 Departamentul	II
1.4 Domeniul de studii de Licență ¹⁾	Medicine
1.5 Ciclul de studii ²⁾	Bachelor
1.6 Programul de studii/ Calificarea	Medicine

2. Date despre disciplină

2.1. Denumirea disciplinei	Clinical Genetics							
2.2 Titularul activităților de curs	Assoc. Prof. Dr. Chiriță Emandi Adela							
2.3 Titularul activităților de laborator	Assoc. Prof. Dr. Chiriță Emandi Adela							
2.4 Anul de studiu	VI	2.5 Semestrul	11	2.6 Tipul de evaluare	Colloquy	2.7 Regimul disciplinei	Conținut ³⁾	SD
							Obligativitate ³⁾	FacD

3. Timpul total estimat (ore pe semestru al activităților didactice)

3.1 Număr de ore pe săptămână	2	3.2 din care: curs	1	3.3 laborator	1
3.4 Total ore din planul de învățământ	28	3.5 din care: curs	14	3.6 laborator	14
Distribuția fondului de timp					ore
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					
Examinări					1
Alte activități					
3.7 Total ore studiu individual					
3.8 Total ore pe semestru		29			
3.9 Numărul de credite⁵⁾					

4. Precondiții (acolo unde este cazul)

4.1 de curriculum	Medical Genetics
4.2 de competențe	Genetic analysis bulletin interpretation

5. Condiții (acolo unde este cazul)

5.1 de desfășurare a cursului	<ul style="list-style-type: none"> • Telephone conversations are not tolerated during the course • Delay of students in the course will not be tolerated, as it proves to be disruptive to the educational process. • Mandatory attendance is required, a maximum of 30% absences being accepted in the course.
5.2 de desfășurare a seminarului/ laboratorului/ proiectului	<ul style="list-style-type: none"> • Telephone conversations are not tolerated during the labs • Delay of students will not be tolerated, as it proves to be disruptive to the educational process. • Mandatory attendance is required in the labs, a maximum of 15% absences being accepted. • Recovery is allowed up to 15% of the total number of paid absences in the last week of the semester (except for medical cases that will require individual Dean's approval).

6. Competențe specifice acumulate

Competențe Profesionale	<ol style="list-style-type: none"> 1. Acquisition of terminology in Clinical Genetics. 2. Ability to describe the clinical picture in main genetic syndromes, correlation with genetic tests. 3. Evaluation of patients with genetic diseases or disorders with genetic component. 4. Developing a testing plan in genetic diseases. 5. Knowledge of the teratogenic effects of prescription drugs; specifying which drugs are allowed/ are prohibited during pregnancy.
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Competențe transversale	1. Preoccupation for professional development by engaging critical thinking skills demonstrated by active participation in the course and laboratory.
	2. Involvement in scientific research activities by participating in the elaboration of papers, studies, specialty articles.
	3. Effective use of information sources and communication and assisted training resources (Internet portals, specialized software applications, databases, on-line courses, etc.) both in Romanian and in an international languages.

7. Obiectivele disciplinei (reieșind din competențele specifice acumulate)

7.1 Obiectivul general al disciplinei	Acquiring the fundamental notions of Clinical Genetics. Genetic diseases are very different, rare and complex, the course's objective is to learn the multidisciplinary approach, work in teams, use of databases and address these diseases to national and/or European networks.
7.2 Obiectivele specifice	<p>Knowledge and understanding of signs and symptoms in genetic pathology, structured on life cycles, correlated with genetic testing for obtaining an accurate diagnosis.</p> <p>Methods of managing these cases from clinical suspicion to the accurate diagnosis, genetic testing, recurrence risk assessment, prenatal diagnosis and treatment, rehabilitation and prevention strategies.</p> <p>Calculating the recurrence risk in single-gene diseases.</p> <p>Differentiation of single-gene diseases from polygenic multifactorial diseases.</p>

8. Conținuturi

8.1 Curs	Metode de predare	Număr de ore	Observații
1. Fundamentals of molecular and cellular human mechanisms of heredity. Types of genetic tests used to diagnose genetic diseases.	INTERACTIVE LECTURE	2	<ul style="list-style-type: none">• Interactive lecture, also presented in Power Point, accompanied by an extremely rich iconography.• Available on University's Moodle e-learning platform• The course is structured according to the European manner of teaching, is updated annually with the latest information from international specialized literature.
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 pathology relevant to the new born and child. 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 component 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, neuro-psychiatric 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	
Bibliografie obligatorie: 1. New Clinical Genetics 3. Andrew Read, Dian Donnai. Scion Publ. Ltd, 2015			
Bibliografie facultativă: 1. Harrison'S Priciples 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 Seminar/ Laborator /stagiul/ proiect	Metode de predare-învățare	Număr de ore	Observații
1. Characteristics of family history in genetic counseling. Identification of different	LECTURE+DEBATE+STUDIES– CASE PRESENTATIONS	2	<ul style="list-style-type: none">• Interactive lecture,

inheritance patterns in the family trees. Establishment of genetic risks. Genetic technologies for diagnosis and research.			also presented in Power Point, available on University's Moodle e-learning platform
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	• Case presentations.
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	Diagnostic algorithms to guide thinking of the future medical practitioner toward targeted investigations necessary to establish a correct diagnosis.
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	• Presentation of investigative methods for the clinical, differential, etiological diagnosis. Ethical principles, support groups for patients.
5. Current recommendations on the supervision of family cancers-prone individuals. Methods of genetic testing in the main types of cancers.		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 the patient, family, collaborators and specialized laboratories. Legal opportunities and social assistance for persons with disabilities.		2	• Verifying the acquisition of the main knowledge by multiple choice questions at the end of the lab.
Bibliografie obligatorii: <ol style="list-style-type: none"> 1. Thompson & Thompson Genetics in Medicine, Nussbaum, McInnes, Willard 8th Ed. Elsevier, 2016 2. Practical Genetic Counselling. Peter S. Harper. Hodder Arnold, 2010 Bibliografie facultativă: <ol style="list-style-type: none"> 1. Smith's Recognizable Patterns of Human Malformation, K. Jones. Saunders, 2013 			

9. Coroborarea conținuturilor disciplinei cu așteptările reprezentanților comunităților epistemice, asociațiilor profesionale și angajatori reprezentativi din domeniul aferent programului

<p>Knowledge of clinical genetics will enable future physicians to identify the genetic causes of the diseases, no matter what 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.</p> <p>Information on pharmacogenomics will allow current students to become highly competent physicians. They will be able to provide clinicians with information about the effect of drugs based on patients genetic testing. Personalized medicine is the medicine of the future. Knowledge of clinical genetics will be an asset in medical practice and career development.</p>

10. Evaluare

Tip activitate	10.1 Criterii de evaluare	10.2 Metode de evaluare	10.3 Pondere din nota finală
10.4 Course	<p><i>For grade 5</i> students must prove knowledge of the main signs in genetic and multifactorial disorders, inheritance patterns, interpreting a family tree</p> <p><i>For grade 10</i>, students must have a thorough knowledge of the genetic and partially genetic disorders, clinical features, positive and differential diagnosis, treatment and prevention options, genetic risk</p>	<p><i>Continuous evaluation</i></p> <p><i>Final evaluation:</i> Exam: written multiple choice test (50 questions, one hour).</p>	<p>20%</p> <p>50%</p>

	assessment, genetic testing methods.		
10.5 Lab	<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> student 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> practical exam</p>	<p>10%</p> <p>20%</p>
<p>10.6 Standard minim de performanță</p> <p>Familiarizing with notions of clinical genetics. Knowledge and understanding of genetic terminology.</p>			

Data completării 04.04.2023	Semnătura titularului de curs Assoc. Prof. Dr. Chiriță Emandi Adela	Semnătura titularului de laborator/stagiu Assoc. Prof. Dr. Chiriță Emandi Adela
Semnătura șefului de disciplină Prof. Dr. Puiu Maria		
Data avizării în departament 4.4.2023	Semnătura directorului de departament Prof. Dr. Cretu Octavian	

Notă:

- 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 – 30 de ore de studiu (activități didactice și studiu individual).
- ⁶⁾ Pentru specializările și/sau disciplinele a căror tematică se regăsește în bibliografia de rezidențiat, aceasta devine obligatorie.