



DISCIPLINE FILE

1. Data about the programme

1.1.	“CAROL DAVILA” UNIVERSITY OF MEDICINE AND PHARMACY
1.2.	FACULTY OF MEDICINE
1.3.	DEPARTMENT 7 Pediatrics
1.4.	DISCIPLINE Medical Genetics
1.5.	DOMAIN OF STUDY: HEALTH – Sectorally regulated within the European Union
1.6.	STUDY CYCLE: LICENCE
1.7.	STUDY PROGRAME: MEDICINE – ENGLISH MODULE

2.2. Data about discipline

2.1.	Name of the discipline in the educational plan: Clinical Genetics in Medical Practice				
2.2.	Discipline code:				
2.3.	Discipline type (FD/SD/CD): CD				
2.4.	Discipline regimen (MD/OPD/):OPD				
2.5.	The holder of the course activities Assoc.Prof.Viorica Elena Radoi				
2.6.	The holder of the seminar activities : Assist. Prof. Monica Stoian				
2.7. Year of study	V	2.8. Semester	II	2.9. Type of evaluation (E/C)	E

2. Total estimated time (hours/semester of didactic activity an self preparation/study

I. Academic training (teaching, practical application, assessment)						
3.1. Nr hours/week	4	From which:	3.2. lecture	2	3.3. seminary/ laboratory	2
3.4. Total hours of educational plan	28	From which:	3.5. lecture	14	3.6. seminary/ laboratory	14
Evaluation (nr. of hours) :						
II. Self preparation/study						
Time allocation						28 hours
Study of course materials, textbooks, books, study of the recommended minimal bibliography						14
Additional research in the library, research through the internet						4
Performing specific activities for preparing projects, laboratories, elaborating reviews or other tasks						4
Specific preparation activities for projects, laboratory work, assignments, and reports						4
Tutoring						
Other activities						2
3.7. Total individual study hours						28

3.9. Total hours per semester (3.4.+ 3.7.)	56
3.10. Number of credits	

3. Preconditions (where applicable)

4.1. of curriculum	Medical Genetics
4.2. of competences	

4. Conditions (where applicable)

5.1. to conduct the lecture	PowerPoint presentations, use of multimedia systems, and projector
5.2. to conduct the seminar / laboratory	Equipped with the necessary apparatus for conducting practical activities

6. Learning outcomes

Knowledge	Skills	Responsibility and autonomy
. Upon completion of the optional course <i>Medical Genetics at the Interface of Specialties</i> , students will acquire an in-depth understanding of the fundamental principles of human genetics and their role in the etiology, diagnosis, and management of hereditary diseases. They will be able to describe the molecular mechanisms involved in the transmission of genetic traits, recognize the importance of genetics in cardiovascular, oncologic, pediatric neurological, and maternal–fetal pathologies, and explain genotype–phenotype correlations. Furthermore, students will gain essential knowledge about modern genetic diagnostic techniques, available genetic tests, and the ethical and	After completing the course, students will demonstrate the ability to apply their theoretical knowledge in clinical or research contexts. They will be able to analyze complex clinical cases involving hereditary genetic disorders, identify the need for specific genetic testing, and collaborate effectively with specialists from related fields such as cardiology, oncology, pediatric neurology, and obstetrics–gynecology. Additionally, students will be capable of interpreting genetic test results, contributing to the differential diagnosis of genetic diseases, and participating in the development of personalized management plans. The acquired knowledge will also enable them to support genetic counseling and prevention activities and to integrate genetic data into evidence-based medical practice.	Students will develop professional competencies and a responsible attitude toward the application of genetic knowledge in everyday medical practice. They will be able to autonomously analyze genetic information relevant to each patient, recognize the limits of their competence, and seek interdisciplinary collaboration when necessary. At the same time, they will display ethical conduct and empathy in their interactions with patients and families, respecting genetic data confidentiality and the principles of informed consent. The skills acquired will enable students to adopt a critical and reflective approach toward the rapid developments in medical genetics, continuously update their knowledge, and contribute responsibly and independently to improving the quality of medical care and genetic counseling.

psychosocial implications of genetic counseling. By assimilating this information, they will be able to integrate theoretical knowledge into a multidisciplinary perspective, recognizing the role of genetics as a bridge between various medical specialties.		

7. Course objectives (aligned with the learning outcomes)

7.1. General objective	The discipline of genetics in clinical practice aims to provide physicians with the theoretical knowledge and practical skills necessary to identify, diagnose, and manage genetic disorders, as well as to counsel patients and their families on genetic risks, treatment options, and prevention.
7.2. Specific objective	Recognition of inheritance patterns: Identify how different genetic diseases are transmitted (autosomal dominant, autosomal recessive, X-linked, mitochondrial). Interpretation of genetic tests: Know how to interpret genetic test results and integrate them into the clinical context. Genetic counseling: Provide patients and families with accurate information about genetic risks, screening options, and prevention. Early diagnosis of genetic disorders: Recognize clinical signs suggestive of genetic diseases and initiate appropriate investigations. Patient management and monitoring: Develop individualized treatment and monitoring plans for diagnosed genetic diseases.

8. Contents

8.1. Lecture	Teaching methods	Observations
1. Medical genetics at the intersection of specialties	PPTX	2h
2. Genetics in hereditary heart disease	PPTX	
3. Genetics in oncology	PPTX	
4. Genetic diseases in pediatric neurology pathology	PPTX	

5. Genetics in maternal-fetal medicine	PPTX	
6. Genetics in maternal-fetal medicine	PPTX	
7. Genetic counseling	PPTX	
8.2. Laboratory/ practical lesson	Teaching methods	Observations
Genetic consultation		2h
Interpretation of genetic tests in hereditary heart diseases - genetic counseling	PPTX+open discussions	2h
3. Interpretation of genetic tests in oncological pathology - genetic counseling	PPTX+open discussions	2h
4. Interpretation of genetic tests in hereditary heart diseases - genetic counseling	PPTX+open discussions	2h
5. Interpretation of genetic tests in NP pathology - genetic counseling	PPTX+open discussions	2h
6. Interpretation of genetic screening and prenatal diagnosis tests - genetic counseling	PPTX+open discussions	2h
7. Genetic diagnosis techniques for various	PPTX+open discussions	2h
Recent bibliography Textbooks and reference works • <i>Harrison's Principles of Internal Medicine</i>: Includes a chapter on "The Practice of Genetics in Clinical Medicine". • <i>Thompson & Thompson Genetics in Medicine</i>: • <i>Oxford Desk Reference: Clinical Genetics</i>: • <i>Principles of Medical Genetics</i> by Gelehrter, Collins, and Ginsburg Online resources and databases • NCBI Bookshelf: Medical Genetics Summaries (MGS) • GeneReviews:		

- [Online Mendelian Inheritance in Man \(OMIM\)](#): A comprehensive database of human genes and genetic disorders.
- [ClinicalTrials.gov](#):

9. Evaluation

Activity type	9.1. Evaluation criteria	9.2. Evaluation methods	9.3. Percentage in the final grade
9.4. Lecture		Exam with 30 questions, each having a single choice answer	80%
9.5. Seminary/ practical activity	Genetic test interpretation" "Genetic counseling"		20%
9.5.1. Individual project (if applicable	-	-	-
9.6. Minimum performance standard 5			

Date of completion :

Signature of the course holder

Signature of the laboratory holder

Date of approval by the
Department Council:

Signature of the Department Director