DISCIPLINE SHEET

ACADEMIC YEAR

2022 - 2023

1. DATA ABOUT THE STUDY PROGRAM

1.1 Institution of higher education	UNIVERSITY OF MEDICINE AND PHARMACY OF CRAIOVA
1.2 Faculty	**MEDICINE**
1.3 Department	I
1.4 Study Domain	HEALTH
1.5 Study cycle	LICENCE
1.6 Study program/ Qualification	**MEDICINE**

2. DATA ABOUT THE DISCIPLINE

2.1 DISCIPLINE NAME				**GENETICS**	
2.2. Discipline code				**MED2205**	
2.3 The holder of course activities				Prof. FLORIN BURADA/Lecturer AMELIA DOBRESCU	
2.4 The holder of seminar activities				Prof. FLORIN BURADA/Lecturer AMELIA DOBRESCU /Teaching Assistant Serban-Sosoi Simona//Teaching Assistant Mihai Gabriel Cucu	
2.5.Academic degree				Professor / Lecturer/Teaching Assistant/Teaching Assistant	
2.6. Employment (base norm/associate)				Base norm/ Base norm /Base norm/ Base norm	
2.7. Year of study	**II**	2.8. Semester	**II**	2.9. Course type (content) 2.10. Regime of discipline (compulsoriness)	**CFD**

3. THE ESTIMATED TOTAL TIME (teaching hours per semester)

3.1 Number of hours per week	**4**	From which - course	**2**	seminary/laboratory	**2**
3.4 Total hours in curriculum	**56**	From which - course	**28**	seminary/laboratory	**28**
Time found distribution (hours)					
Study from manual, course support, bibliography, and notes					**14**
Additional documentation in the library, specialized electronic platforms and, on the field					**7**
Training seminars / labs, homework, reports, portfolios, and essays					**12**
Tutoring					**2**
Examinations					**3**
Other activities, counselling, student scientific programs					**6**

3.7 Total hours of individual study	**44**
3.9 Total hours per semester	**100**
3.10 Number of credits	**4**

4. PREREQUISITES (where appropriate)

4.1 curriculum	Acquaintances of Cellular and Molecular Biology, Biochemistry, Physiology, Embryology.
4.2 competency	

5. CONDITIONS (where appropriate)

5.1. of curse deployment	Lecture Hall with projector
5.2. of seminary/ lab deployment	Genetics Lab Individual study of the theoretical and practical notions

6. SPECIFIC COMPETENCES ACCRUED

PROFESSIONAL COMPETENCES

C1. Identification the disease status and establishing the correct diagnosis.
C3. Correct assessment of disease risk and context of occurrence of an individual / collective disease, followed by the selection and application of appropriate prophylaxis measures.
C4 – To address health issues/illness from the perspective of community specifics, directly related to the social, economic and/or the cultural specificity.
C5. To initiate and conduct a scientific research activity and / or a training activity inside the field of competence

TRANSVERSAL COMPETENCES	**CT1**. Autonomy and responsibility • the acquisition of moral reference points, the formation of professional and civic attitudes, that will allow to the students to be fair, honest, helpful, understanding, unconflictuals,to cooperate and to be comprehensive in the face of suffering, to be available to help people, and to be interested in community development; • to know, to respect and to contribute to the development of moral values and professional ethics; • to learn how to recognize the problems when they arise ,and provide solutions for solving them. **CT2**. Social interaction • to recognize and to have respect for diversity and multiculturalism; • to have or to learn how to develop teamwork skills; • to communicate orally and in writing the manner of work requirements, the obtained results , to consult with the team; • to engage themselves in voluntary activities, to know the essential problems of the community. **CT3.** Personal and professional development • to have opening to lifelong learning, • to be aware for self-study as a basis of personal autonomy and professional development; • to derive the optimum and creative potential in their own collective activities; • to know how to use information and communication technologies.

7. DISCIPLINE OBJECTIVES (based on the grid of specific competences acquired)

7.1 The general objective of the discipline	Discipline of Genetics is aimed at second year students with the overall objective of understanding the heredity and variability of human beings. In-depth knowledge of genetic mechanisms is particularly important for understanding of many aspects of the etiopathogenesis of hereditary and multifactorial diseases, and for the accurate interpretation of the various phenotypic traits and clinical forms of these diseases. The study of the relationship between heredity and disease provides a new perspective on modern personalized medicine.
7.2 The specific objectives of the discipline	Upon completion of discipline the student will be able to acquire: **Theoretical knowledges:** - recognize features in a patient's medical history, physical examination or investigations that suggest the presence of genetic disease; - know clinical features of the most common monogenic and chromosomal disorders - recognize patterns of inheritance of genetic diseases - distinguish genetic factors associated with susceptibility to cancer - recognize the contribution of genetic and environmental factors in multifactorial diseases, - describe appropriate genetic techniques and approaches used in genetic diagnosis and carriers detection - know the existence of and justification for screening programs to detect genetic disease - know major ethical issues - estimate and communicate the recurrence risks concept in a appropriate manner - communicate genetic information in a clear and non-directive manner that is suitable for individual **Technical skills** : - identify the appropriate genetic methods used for diagnosis of genetic diseases - interpret the results of common cytogenetic, molecular genetic, and biochemical genetic diagnostic techniques efficiently; - estimate recurrence risks for mendelian and multifactorial disorders in affected families; - recognize patterns of inheritance of genetic diseases, - identify the patients with genetic disease predisposition and predictive tests that are appropriate for the condition in their family and advise patients of the benefits, limitations, and risks of genetic tests; - effectively use resources such as medical textbooks, research articles, and

	computer-based systems to obtain current and appropriate information necessary for good patient care; **Attitudes:** - recognize the importance of patient confidentiality; - utilize community support services and agencies, in particular, support groups for genetic diseases, appropriately - respect patients' religious, cultural, social, and ethical beliefs - cope emotionally with patient responses; - recognize the limitations of their own skills and seek consultation when necessary; - respect the patients' rights - apply ethical principles related to medical practice - recognize the signs of abuse or mistreatment of patients and inform the competent legal courts - acquire and use proper of medical informations - communicate in appropriate manner with patient and family members - work with other specialists for a multidisciplinary team approach

8. CONTENTS

8.1 Course (content units)	22 hours
C1. INTRODUCTION IN MEDICAL GENETICS. Definition and objectives of medical genetics. **DNA – CARRIER OF GENETIC INFORMATION.** Primary structure of nucleic acids. Secondary structure of DNA. Physical properties of DNA. Organization of DNA within the nucleus. Chromatin. Morphogenesis of metaphasic chromosomes.	**2 h**
C2. HUMAN GENOME. STRUCTURE OF GENE. DNA REPLICATION. Structure of the human genome. Gene structure. Enzymes involved in DNA replication. Mechanism of DNA replication.	**2 h**
C3. EXPRESSION OF GENETIC INFORMATION. Transcription. Pre-mRNA processing. Translation. Regulation of gene expression	**2 h**
C4. GENETIC VARIABILITY. GENIC MUTATIONS. DNA REPAIR MECHANISMS. Genetic recombination. Types of genetic mutations. Gene mutations: substitution, deletion, insertion. DNA repair pathways. DNA repair-deficiency disorders.	**2 h**
C5. MONOGENIC INHERITANCE. Mendelian laws of heredity. Characteristic of monogenic inheritance Autosomal dominant inheritance. Autosomal recessive inheritance. Recessive X-linked inheritance. Dominant X-linked inheritance.	**2 h**
C6. MONOGENIC AUTOSOMAL DISORDERS. Autosomal dominant disorders: Marfan Syndrome, Achondroplasia, Neurofibromatosis type I, Familial hypercholesterolemia, Osteogenesis imperfecta. Autosomal dominant polycystic kidney disease. Autosomal recessive disorders: Phenylketonuria, Albinism, Cystic fibrosis, Sicklemia, Hemochromatosis, Lysosomal storage diseases.	**2 h**
C7. MONOGENIC SEX-LINKED DISORDERS. X-linked recessive diseases: Duchenne muscular dystrophy, Becker's muscular dystrophy, hemophilia A, hemophilia B, Red/green colour blindness. X-linked dominant diseases: X-linked hypophosphatemia, Incontinentia pigmenti, Rett Syndrome.	**2 h**
C8. CHROMOSOMAL DISEASES. Autosomal diseases: Down Syndrome (trisomy 21), Patau Syndrome (trisomy 13), Edwards Syndrome (trisomy 18). Sexual chromosome diseases: Turner Syndrome (monosomy X), Klinefelter Syndrome (trisomy XXY), Trisomy X (Triple X Syndrome), Trisomy XYY	**2 h**
C9. CHROMOSOMAL DELETIONS AND MICRODELETION SYNDROMES NON-CLASSIC MONOGENIC INHERITANCE: UNIPARENTAL DISOMY. GENOMIC IMPRINTING. Deleetion and microdeletion syndromes: Cri du chat syndrome, Wolf-Hirschhorn syndrome, velo-cardio-facial/DiGeorge syndrome, Williams syndrome. Disorders associated with uniparental disomy/genomic imprinting: Beckwith-Wiedemann syndrome, Prader-Willi syndrome, Angelman syndrome.	**2 h**
C10. DYNAMIC MUTATIONS. Characteristics of dynamic mutations. Genetic diseases caused by dynamic mutations: Fragile X Syndrome (fra-X), Huntington's disease, Myotonic dystrophy Dynamic mutations. **GENETIC HETEROGENEITY.** Locus heterogeneity, Allelic heterogeneity, Clinical heterogeneity.	**2 h**
C11. MITOCHONDRIAL GENOME. Mitochondrial genome. DNA mt replication. Mitochondrial transcription and translation. Mitochondrial mutations and pathology. Mitochondrial diseases.	**2 h**
C12. MULTIFACTORIAL INHERITANCE. MULTIFACTORIAL DISEASES. Characteristics. Multifactorial diseases. **CONGENITAL ANOMALIES.** Classification of the congenital abnormalities. Causes of congenital anomalies.	**2 h**
C13. NORMAL SEXUAL DIFFERENTIATION. DISORDERS OF SEX DEVELOPMENT. Genetics of sexual development. Disorders of sexual development.	**2 h**
C14. CANCER. Cancer genes. Hereditary cancers.	**2 h**

BIBLIOGRAPHY 1. The taught course. 2. Mircea Covic, Dragos Stefanescu, Ionel Sandovici, Gorduva EV. Medical Genetics, 3th Edition, Polirom Publishing, 2017 3. Nussbaum R, McInnes R, Willard H. *Thompson & Thompson Genetics in Medicine,* 8th Edition, Elsevier, 2015. Romanian Edition, Ed.Hipocrate 2018. 4. Turnpenny P, Ellard S. Emery's Elements of Medical Genetics, 15th Edition, Elsevier, 2017	
8.2 Practical work (topics / themes)	**28 h**
T 1. Morphology of human chromosomes; morphologic and specific types	**2 h**
T 2. Analysis of human chromosomes	**2 h**
T 3. Cell cycle. Mitosis	**2 h**
T 4. Meiosis	**2 h**
T 5. Chromosome banding.	**2 h**
T 6. Normal human karyotype.	**2 h**
T 7. Numerical chromosome abnormalities	**2 h**
T 8. Structural chromosome abnormalities	**2 h**
T 9. Inheritance pattern of normal and pathological monogenic features.	**2 h**
T 10. Analysis of the family pedigree – autosomal inheritance	**2 h**
T 11. Analysis of the family pedigree – sex-linked inheritance	**2 h**
T 12. Molecular genetics techniques in diagnosis of genetic diseases	**2 h**
T 13. Prenatal screening and diagnosis	**2 h**
T 14. Genetic counseling. Recoveries	**2 h**
BIBLIOGRAPHY 1.The discipline protocols. 2. F. Burada. Principles and applications in medical genetics. Sitech Publishing, 2018. 3. Mircea Covic, Dragos Stefanescu, Ionel Sandovici, Gorduva EV. Medical Genetics, 3th Edition, Polirom Publishing, 2017 4. Nussbaum R, McInnes R, Willard H. *Thompson & Thompson Genetics in Medicine,* 8th Edition, Elsevier, 2015. ediția în limba română, Ed.Hipocrate 2018. 5. Turnpenny P, Ellard S. Emery's Elements of Medical Genetics, 15th Edition, Elsevier, 2017	

9. CORROBORATING THE DISCIPLINE CONTENT WITH THE EXPECTATIONS OF EPISTEMIC COMMUNITY REPRESENTATIVES, PROFESSIONAL ASSOCIATIONS AND EMPLOYEE REPRESENTATIVES RELATING TO THIS PROGRAM

Discipline of Genetics is a fundamental and applied science, which has a major role in the theory and practice of medicine.
Discipline of Genetics has a clinical component consisting in the study of the relationship between heredity and disease.
Discipline of Genetics has also a medico-social component as genetic diseases have become a major public health problem.

10. MHETODOLOGICAL LANDMARKS

Types of activity	Techniques of teaching / learning, materials, resources: lecture session, interactive course, learning through questions. In case of special situations (alert states, emergency states, other types of situations that limit the physical presence of people) the activity can be carried out online using computer platforms approved by the faculty / university. The online education process will be adapted accordingly to ensure the fulfillment of all the objectives set out in the discipline sheet.
Course	There are used the next combined methods: lecture session, heuristic conversation, discussion.
Practical work	There are used the next combined methods: demonstration, discussion, experiment.
Individual study	Study after the course support, study of the laboratory work, additional documentation in the library, references study.

11. RECOVERY PROGRAM

	No. absences that can recover	**Location of deployment**	**Period**	**In charge**	**Scheduling of topics**
Absences recoveries	3	Genetics Lab	The last week of the semester	The teacher who performs the practical work	According to the internal schedule
Schedule consultations /	2 hrs/week	Genetics Lab	Weekly	All teaching assistants	The theme of the week

Students' Scientific Program					
Program for students poorly trained	2 hrs/week	Genetics Lab	Weekly	All teaching assistants	The theme of the week

12. ASSESMENT

Activity	**Types of assesment**	**Methos of evaluation**	**Percentage from final grade**
Lecture	Formative assessment through surveys during the semester Summative assesment during the exam	Written exam	70%
Practical work	Formative assesment through surveys during the semester. Periodic assesment during the semester Summative assesment during the exam	Written examff	25%
Periodic assesment			
Assesment of individual activity			5%
Minimum performance standard			At least 50% for each component of the evaluation

13. GUIDANCE AND COUNSELLING PROGRAMS

Professional guidance and counselling programs (2 hours/monthly)

Scheduling the hours	**Location**	**In charge**
Last Friday of each month	Genetics Lab	Prof. Florin Burada Lecturer Amelia Dobrescu

Endorsement date in the department: 27.09.2022

Department Director,	**Coordinator of study program,**	**Discipline holder,**
Prof. Ion Mindrila	**Prof. Marius Eugen Ciurea**	**Prof. Florin Burada**