SUBJECT OUTLINE

1. Programme of study description

1.1.	**THE "CAROL DAVILA" UNIVERSITY OF MEDICINE AND PHARMACY**
1.2.	**THE FACULTY OF MEDICINE / THE CLINICAL DEPARTMENT**
1.3.	**DISCIPLINE**
1.4.	**DOMAIN OF STUDY: Healthcare – regulated sector within the EU**
1.5.	**CYCLE OF STUDIES: BACHELOR'S DEGREE**
1.6.	**PROGRAMME OF STUDY: MEDICINE**

2. Subject description

2.1.	**Name of the subject/compulsory subject/elective subject within the discipline:**
2.2.	**Location of the discipline: Department of Medical Genetics**
2.3.	**Course tenured coordinator:** Prof. Bohiltea Laurentiu Camil, MD, PhD Lect. Viorica Elena Radoi, MD, PhD Lect. Radu-Ioan Ursu, MD, PhD
2.4.	**Practicals/clinical rotations tenured coordinator:** Lect. Radoi Viorica Elena, MD, PhD Lect. Ursu Radu Ioan, ND, PhD TA Paraschiv Cerasela, MD, PhD TA Zeleniuc Monica, MD, PhD TA Raicu Florina, Biol, PhD TA Grozescu Stefan Traian, MD, PhD TA Leonte Dordia Laura, MD, PhD TA Dragotoiu Rodica, MD, PhD TA Focsa Ina, MD (hourly payment) TA Sabau Delia, MD (hourly payment)

2.5. Year of study	1	**2.6. Semester**	2	**2.7. Type of assessment**	Written Exam, Practical Exam	**2.8. Subject classification**	Mandatory

3. Total estimated time (hours/semester of didactic activity) – teaching module

Number of hours per week	5	**Out of which: course**	2	**Clinical rotation**	3
Total number of hours from curriculum	70	**Out of which: course**	**28**	**Clinical rotation**	42
Distribution of allotted time					**Hours**
Study from textbooks, courses, bibliography, and student notes					
Additional library study, study on specialized online platforms and field study					
Preparing seminars / laboratories, assignments, reports, portfolios and essays					
Tutoring					
Examinations					
Other activities					
Total hours of individual study					
Number of credit points	3				

4. Prerequisites (where applicable)

4.1. of curriculum	Not necessary
4.2. of competencies	Not necessary

5. Requirements (where applicable)

5.1. for delivering the course	
5.2. for delivering the clinical rotation	

6. Acquired specific competencies

Professional competencies (expressed through knowledge and skills)	Personelle with a great experience in the field of medical genetics: genetic departments within hospitals, clinics and private laboratories, with an expertise in genetic testing, genetic counselling, in cytogenetics, molecular genetics and clinical genetics
Transversal competencies (of role, of professional and personal development)	Identification of roles and responsibilities within the team, application of communication techniques and effective team work Efficient use of information sources and communication resources and assisted professional training.

7. Subject learning objectives (based on the scale of acquired specific competencies)

7.1. General learning objective	Acquiring the fundamental medical genetics knowledge
7.2. Specific learning objectives	The recognition and approach of the genetic factor in the realization of human pathology. Knowledge on the principles of cytogenetic analysis techniques (karyotype) and molecular cytogenetics (FISH testing and comparative genomic hybridization) Knowledge on the principles of PCR-based techniques. DNA sequencing. Principle, DNA sequencing by Sanger method, stages of sequencing technique, interpretation, utility of DNA sequencing. The ability to evaluate and interpret the results of genetic investigations in a clinical context. The value of applying cytogenetics, molecular cytogenetics and PCR-based methodologies for diagnostic purposes, establishing prognosis and monitoring, in different clinical fields. Understanding the mechanisms of action at the cellular and molecular level with applications in medical practice Designing and applying a management plan suitable for the identified genetic condition Acquisition of genetic counseling skills Acquiring the skills to establish optimal communication relationships between the geneticist and the patient/family

8. Content

8.1. Course	**Teaching methods**	**Observations**
Course 1	Medical Genetics – Introduction	The lectures are taught in the amphitheater which is technically
Course 2	Nucleic Acids Structure. Nucleic and mitochondrial DNA	
Course 3	DNA Replication	

Course 4	Human Chromosomes - Structure and functions	equipped for this purpose - laptop, video projector All courses are also on electronic support and are permanently updated according to the treatises and specialized journals and the information published online in genetic pathology The department has a library and the possibility of online access to obtain the necessary information.
Course 5	Genes - structure and functions	
Course 6	Protein Synthesis	
Course 7	Mutations	
Course 8	Mutation Repair Systems	
Course 9	Genetic disorders - chromosomal and monogenic	
Course 10	Multifactorial disorders. Epigenetic pathology	
Course 11	Oncogenetics	
Course 12	Genetic disorders prophylaxis	
Course 13	Bioethics in medical genetics	

8.2. Clinical rotation	**Teaching methods**	**Observations**
CR 1	Meiosis; The cell cycle: Meiosis I/Meiosis II (interphase, prophase, metaphase, anaphase, telophase), Crossing-over phenomenon, Genetic recombination through crossing-over, Consequences of meiosis, Nondisjunctions (definition, causes of nondisjunction, mechanisms)	All practical classes are also on electronic support and are permanently updated according to the treatises and specialized journals and the information published online in medical genetic pathology. The department has a genetics laboratory. Optical microscopes are made available to students. The department has a library and the possibility of online access to obtain the necessary information
CR 2	Formation of the gametes (Spermatogenesis/Oogenesis). Anomalies of female and male gametogenesis	
CR 3	Sex chromatin: X-chromosome inactivation (Barr body, X-chromatin, X-chromosome inactivation scheme, Exceptions to inactivation, X-chromosome inactivation profile); Y chromatin (F corpuscle)	
CR 4	Normal and pathological karyotype: Introduction, Chromosome structure, Chromosome number, Chromosome nomenclature, Chromosome analysis methods (Preparation of metaphase chromosomes, Specific identification of metaphase chromosomes); Chromosome banding methods - G banding (conventional), C banding, Q banding, R banding (reverse G banding / R banding by fluorescence using Acridine Orange), T banding, NOR banding, DAPI banding. Fluorescence in Situ Hybridization	

CR 5	Study of chromosomal aberrations: Numerical aberrations (Aneuploidy: monosomy, nullisomy, trisomy, tetrasomy/ Polyploidy: triploidy, tetraploidy); Structural aberrations (deletions, insertions, inversions, duplications, ring chromosomes, isochromosomes, translocations)/ Other aberrations (mixoploidy) (mosaics, chimeras)
CR 6	Patterns of inheritance of normal and pathological characters (part I): Mendel's laws, Segregation of Mendelian traits, Independent distribution of two different characters, Segregation of parental genotypes (Autosomal dominant/recessive inheritance, X-linked dominant/recessive and Y-linked inheritance patterns, mitochondrial inheritance), Autosomal dominant inheritance (criteria, traits, examples clinical conditions - Huntington's disease, Marfan syndrome), Autosomal recessive inheritance (criteria, features, clinical examples - Galactosemia, Homocystinuria, Cystic fibrosis), X-linked recessive inheritance (criteria, features, clinical examples - Duchenne muscular dystrophy, X-fragile), X-linked dominant inheritance (criteria, traits, clinical examples – Vitamin-D resistant rickets), Y-linked inheritance (criteria, traits, clinical examples – Hypertrichosis of the ear), Mitochondrial inheritance (criteria, traits, examples clinical – Kearns-Sayre syndrome, Leber hereditary optic neuropathy)
CR 7	Patterns of inheritance of normal and pathological characters (part II)
CR 8	Hardy-Weinberg Law: Factors that disturb the Hardy-Weinberg equilibrium (inbreeding, selection, mutations, genetic drift/ human isolates, migration); Applications of the Hardy-Weinberg law (gene load, estimation of mutation frequencies, gene size, carrier frequencies)
CR 9	Genetic pedigree: Definition, Symbols and method of creating a pedigree, Family investigation.
CR 10	Genetic consultation and counseling: Indications for genetic counseling, Information in genetic counseling, Bayes'

	Theorem, Genetic counseling/counseling process, Genetic screening, Prenatal diagnosis of genetic diseases (Amniocentesis, Chorionic villus biopsy, Fetal blood analysis, Maternal serum screening – bi/triple test, Applications of DNA analysis in prenatal diagnosis)	
CR 11	Erythrocyte genetic systems, plasma: Hemoglobin (hemoglobin structure, expression of various types of hemoglobin, globin chain structure, synthesis and control of hemoglobin expression, diseases affecting hemoglobin structure, diseases affecting hemoglobin synthesis), ABO blood group system (ABO antigens, hemolytic disease of the newborn, Bombay phenotype, subgroups A1 and A2), Rh system (Rh antigens, RH genotypes/phenotypes, heredity, Rh hemolytic disease of the newborn), Lewis system, Duffy system, G6PD system, PAE system	
CR 12	Molecular genetics techniques used in the diagnosis of genetic diseases (part I): I). Comparative genomic hybridization II). Gene analysis by DNA amplification technique: - PCR - electrophoresis of nucleic acids - restriction enzymes - DNA sequencing - RT-PCR	
CR 13	Molecular genetics techniques used in the diagnosis of genetic diseases (part II): III). Polymorphism analysis techniques: - RFLPs (restriction fragment length polymorphism) - AFLP (amplified fragment length polymorphism) IV). DNA and RNA hybridization: - principle - Southern blotting - Northern blotting V). Genomic banks: - construction of a genomic DNA bank - construction of a cDNA bank	
CR 14	Practical exam	

Bibliography for course and clinical rotation

9. Corroboration of the subject content with the expectations of the representatives of the epistemic community, professional associations, and major employers in the field of the programme of study

The professional training of the student in the Medical Genetics Discipline aims to deepen the genetic aspects in medical pathology, the recognition of genetic diseases and the organization of an adequate management plan for genetic diseases. In addition, the training of the future doctor is aimed at a proper professional communication with the future employer.

10. Assessment

Type of activity	**Assessment criteria**	**Assessment methods**	**Assessment weighting within the final grade**
Course	Grid system – 50 questions of 1 point each (maximum score 50 points): 45.1 – 50 points: grade 10 40.1 – 45 points: grade 9 35.1 – 40 points: grade 8 30.1 – 35 points: grade 7 25.1 – 30 points: grade 6 20.1 – 25 points: grade 5 Below 20 points: not promoted	Grid system 50 questions - 4 different grid options, each question with 4 different answer options: - 30 simple choice type questions (1 correct answer only) - 20 multiple choice type questions (2, 3 or 4 correct answers)	75%
Clinical rotation	Interpretation of a karyogram Interpretation of a result - molecular test Creating a family tree Providing appropriate genetic counseling	Oral/written assessment (grid/written - synthesis system)	25%
Minimum performance standard			
Grade 5			

Date of filing **Signature of the course tenured coordinator** **Signature of the seminar tenured coordinator**

Date of approval in the Council of the Department: **Signature of the Head of the Department**