DISCIPLINE GRID

1. Programme:

1.1.	**CAROL DAVILA UNIVERSITY OF MEDICINE AND PHARMACY BUCHAREST**
1.2.	**FACULTY OF DENTISTRY / 1st DEPARTMENT**
1.3.	DIVISION: **GENETICS**
1.4.	STUDY DOMAIN: **Health, sectoral regulated within European Union**
1.5.	STUDY LEVEL: **LICENCE**
1.6.	STUDY PROGRAMME: **DENTAL MEDICINE IN ENGLISH**

2. Discipline:

2.1.	DISCIPLINE NAME: **GENETICS**						
2.2.	LOCATION: **Bucharest, 19-21, Dimitrie Gerota Street, 1st floor**						
2.3.	Lectures tenure: **Emilia Severin – Professor**						
2.4.	Practical classes tenure: **Crenguța Albu – Assoc.Prof**						
2.5. Study year	**II**	**2.6.** Semester	**IV**	**2.7.** Evaluation	**Exam**	**2.8.** Type of discipline	**CD/FD**

3. Estimated total time (hours/semester)

No. hours/week	**4**	out of which	**Lectures: 2**	**Laboratory session: 2**
Total hours out of learning schedule	**56**	out of which	**Lectures: 28**	**Laboratory sessions: 28**

Time distribution	**hours**
Textbook study, lecture support, bibliography and notes	**28**
Supplementary documentation activity in the library, on online platforms	**7**
Practical activity support material, homework, portfolio and essays	**7**
Tutorial activity	**1**
Examinations	**1**
Other activities	**-**
Total hours of individual study	**44**
Total hours per semester	**100**
Credits	**4**

4. Preconditions

4.1. curriculum	Knowledge aquired through the courses taken in the year: - Biochemistry - Molecular Biology - Embryology
4.2. proficiencies	Knowledge of optical microscope working techniques.

5. Conditions

5.1. for lecture activity	Auditorium with a minimum of 125 seats, equipped with specific technical amenities (computer/laptop, projector, whiteboard, PowerPoint or PDF course material, teaching aids, etc.). Students are expected to adhere to university education norms, including attendance, punctuality, and refraining from using mobile phones. Student tardiness during lectures and seminars/laboratory sessions is found to be disruptive to the educational process. Active and interactive learning is encouraged.
5.2. for laboratory activity	Laboratory equipped with a video projection system, whiteboard, microscopes, teaching materials, reagents, and instruments specific to genetic investigations. Active participation in practical classes. Requests for postponement (absence) from practical classes will only be accepted for valid reasons. Practical and applied learning.

6. Accumulated skills

6.1. Proficiencies ***(knowledge and abilities)***	**I. Knowledge (cognitive dimension)** ● Understanding and correct use of terminology, concepts, and fundamental principles of human and medical genetics and genomics. ● Demonstrating analytical knowledge/interpretation capacity of genetic and genomic concepts. ● Integrating this knowledge into modern precision medicine: predictive, personalized, preventive, and participatory. Specifically, ● Understanding the principles of chromosomal analysis methods and performing a chromosomal test with the ability to interpret its results. ● Specifying the diagnosis and genetic counseling for chromosomal syndromes with oro-dento-craniofacial involvement (Down, Turner, Klinefelter syndromes). ● Creating a family and maternal-fetal pedigree based on family and maternal-fetal history, with the ability to interpret the mode of transmission of a pathological trait. ● Understanding and applying the principles and indications of genetic consultation. ● Evaluating the risk of disease. ● Recognizing (identifying) appropriate prenatal screening methods and practical indications for prenatal diagnosis. **II. Abilities (functional dimension)** ● Identification and utilization of optimal genetic tests based on the cause of the disease or anomaly under investigation. ● Laboratory work, both in teams and individually. ● Communication with the patient. ● Proper skills and attitudes in addressing medical and bioethical problem cases. ● Creativity and innovation in scientific research.
6.2. Transversal skills ***(role, professional and personal***	**III. Role skills** ● Autonomy / responsibility / social interaction ● Identifying objectives to be achieved, available resources, completion conditions, work stages, work schedules, associated deadlines, and related risks.

development)	● Familiarization with the roles and specific tasks within a teamwork context, as well as task delegation for subordinate levels. ● Recognizing roles and responsibilities within a multidisciplinary team and applying efficient communication and collaboration techniques within the team. ● Applying professional ethics principles and norms. **IV. Professional and personal development skills** Continuous personal and professional development: ● Awareness of the need for continuous training. ● Efficient use of informational sources and assisted professional communication and training resources (Internet portals, specialized software applications, databases, online courses, artificial intelligence, etc.) both in Romanian and in an international language of communication.

7. Objectives **(based on the grid of acquired specific skills)**

7.1. General Objective	GO1.Acquisition by second-year students of fundamental knowledge in the field of structural, functional genetics, and genetic engineering with a major role in the current and prospective medical theory and practice. GO2. Integration of this knowledge into modern medicine: prospective, predictive, personalized, preventive, and participatory.
7.2. Specific Objectives	SO1.. The objectives include the formation of a knowledge base that allows for understanding and addressing issues related to: ● Human heredity and normal/pathological variability, genetic disease diagnosis, genetic counselling, and genetic therapy. ● The genotype-phenotype relationship and the involvement of genetic factors in the occurrence of abnormalities and diseases with oro-dento-craniofacial localization. SO2. The development of skills and abilities necessary for applying genetic investigation concepts and methods (laboratory techniques: cytogenetic tests, pre- and postnatal genetic screening and diagnosis) in medical practice and providing genetic counselling/personalized care to patients with genetic conditions. SO3. Training in laboratory work, both as part of a team and individually.

8. Content

8.1. Lectures	No. hrs/topic	Teaching method	Obs.
Course 1: **Introduction to Human Genetics: Terminology, Concepts, and General Principles.** 1.1 The Content of Human Genetics: Genetics, the science of heredity and variability; Human genetics, a fundamental, clinical, and socio-medical discipline. 1.2 Genetic and Biological Individuality 1.3 Determinism of Human Traits 1.4 The Genotype-Phenotype-Environment Relationship 1.5 The Impact of Human/Medical Genetics on Daily Life 1.6 The Molecular Basis of Human Heredity: Nucleic Acids and Genetic Information	2	Interactive presentation of the material according to the curriculum, using multimedia tools, PowerPoint presentations, and educational videos.	
Course 2: The Structure and Functions of the DNA Molecule 2.1 Discovery of the Molecular Structure of DNA 2.2 Characterization of the DNA Molecule's Structure 2.3 Alternative Structures of the DNA Molecule 2.4 Characteristics of the Human DNA Molecule (Nuclear and Mitochondrial) 2.5 The relationships between the structure and functions of DNA.	2		
Course 3: Nuclear and mitochondrial DNA 3.1 Nuclear DNA and Mitochondrial DNA. 3.2 Cellular RNAs: Structure, Types, Functions. 3.3 Nucleic acid analysis: molecular hybridization, recombinant DNA, DNA sequencing – applications in medicine. 3.4 DNA Organization in Eukaryotic Cells: Genetic Apparatus, Nuclear Chromatin.	2		
Course 4: The Chromatin and the mitotic Chromosome in Eukaryotes 4.1 Euchromatin and Heterochromatin 4.2 Chromosome Morphology 4.3 A Brief Description of the Human Karyotype 4.4 Heteromorphism of Human Chromosomes 4.5 Chromosomal Analysis Techniques and Indications for Chromosomal Analysis	2		
Course 5: Human Chromosomes: Gonosomes 5.1 Sex Chromosomes and Sex Determination 5.2Sexual Determinism and Sexual Differentiation 5.3 Disorders of Sexual Development 5.4 Conclusions: Chromosomal Abnormalities and Their Phenotypic Effects in Humans	2		
Course 6: Replication of Nuclear and Mitochondrial DNA. 6.1 How is DNA Replicated? - Messelson & Stahl Experiment 6.2 DNA Replication in vitro / in vivo 6.3 Replication of Nuclear Chromosomes and the mt Chromosome 6.4 Replication and DNA distribution during cell division	2		
Course 7: The Gene - Classical vs. Modern Concepts 7.1 Genes - Structure, Analysis, and Gene Localization 7.2 The Chromosomal Theory of Inheritance	2		

7.3 Current Concepts of Gene Structure and Localization in Chromosomes 7.4 Characteristics of Human Genes / Genes Involved in the Development of the Craniofacial Complex 7.5 The Human Genome			
Course 8: Gene Interactions 8.1 Allelic / Non-allelic Interactions 8.2 Pleiotropy 8.3 Genetic Heterogeneity 8.4 Phenocopy	2		
Course 9: Gene Expression (Gene Function) 9.1 Transcription 9.2 The Genetic Code 9.3 Translation 9.4 Regulation of Gene Expression	2		
Course 10: Reverse Transcription; Human Genetic Diseases and Defects 10.1 Structure and Functions of the Viral Genome / Replicative Cycle 10.2 Chromosomal Disorders (Characteristics) 10.3 Nuclear Monogenic Diseases (Mendelian Diseases) 10.4 Exceptions to Mendelian Inheritance	2		
Course 11: Human Genetic Diseases and Defects 11.1 Multifactorial Human Diseases 11.2 Diseases Caused by Pathogenic Variants in Somatic Genes 11.3 Carcinogenesis, Malignant Phenotype, and Genetic Predisposition in Cancer, Hereditary Cancers 11.4 Oncogenes vs. Proto-oncogenes, Tumor Growth Suppressor Genes, Stability Genes, MicroRNA Genes.	2		
Course 12: Human Genetic Variability 12.1 Individual Variations 12.2 Genetic Variation and DNA Profiling 12.3 Sources of Genetic Variation: Mutation, Genetic Recombination, and Migrations 12.4 Phenotypic Effects of Benign / Pathogenic / Common / Rare Gene Variants on Oral Health	2		
Course 13: Developmental Genetics and Human Congenital Abnormalities 13.1Embryofetal Developmental Abnormalities: Definition, Classification, Prevalence 13.2 Causes of Congenital Abnormalities 13.3 Morphogenesis Errors 13.4 Teratogenesis	2		
Course 14: Ecogenetics/Pharmacogenetics/ Pharmacogenomics 14.1 A Brief Chronology of Pharmacogenetics 14.2 Possible Effects of a Medication / Pharmacogenetic "Accidents" 14.3 Pharmacogenetic / Pharmacogenomic Testing 14.4 **FINAL CONCLUSIONS OF THE GENETICS COURSE**	2		

8.2 Laboratory Sessions	**No. hrs/topic**	**Teaching method**	**Obs.**
Human Cytogenetics	2		

Practical Work (P.W.) 1 - Human Chromosomes and Normal Karyotype		Structured by the type of laboratory activity: ● Active-participatory methods, heuristic conversation, using multimedia tools within the student group. ● Observation, practical demonstration, applied exercises, experiments, case studies applied in groups or individually. ● Preparation of reports/projects . ● Practical applications based on theoretical concepts and principles (biological samples, genetic investigation techniques, presentations of analysis reports, and interpretation of results with students). ● PowerPoint presentations, educational videos.	
Human Cytogenetics **P.W.2** - Cell Cycle, Meiotic Chromosomes, and Meiosis in Humans	2		
Human Cytogenetics **P.W.3** - Chromosomal Aberrations: Types, Mechanisms, Examples.	2		
Human Cytogenetics **P.W.4** - Chromosomal Pathology with Craniofacial Involvement	2		
Human Cytogenetics **P.W.5** - Sex Chromosomes and Sexual Chromatin	2		
Human Monogenic Heredity **P.W.6 -** Mendelian Laws of Inheritance – Practical Applications	2		
Human Monogenic Heredity **P.W.7 -** Models of Monogenic Trait Transmission / Pedigree Method (Composition and Interpretation), Family Studies	2		
Human Monogenic Heredity **P.W.8 -** Simple Human Traits I (Blood Group Genetics, Hemoglobin)	2		
Human Heredity **P.W.9 -** Simple Human Traits I (Taste Sensitivity, PTC, Salivary Secretor Status)	2		
Human Polygenic Heredity **P.W.10** - Quantitative Human Traits (Cephalometrics in Dental Medicine – Constitutional Types in Genetic Pathology)	2		
Human Polygenic Heredity **P.W.11** - Quantitative Human Traits (Identity and Individuality, Dermatoglyphics in Medical Genetics)	2		
Integration of Genetic Knowledge into Medical Practice **P.W.12** - Genetic Consultation and Genetic Counseling / Prenatal Screening and Diagnosis	2		
Human Population Evolution **P.W.13** - Human Population Genetics – Hardy-Weinberg Law (Applications and Problems)	2		
P.W.14 - Final Conclusions – Consultations - Make-up Sessions	2		

8.3. Bibliography for lectures and laboratory/practical sessions

REQUIRED READING FOR STUDENTS:

1. Taught Course – 2024, Severin Emilia
2. The PowerPoint presentations discussed during the practical sessions. - 2024, Severin E, Albu C
3. Strachan T, Lucassen A (2023) – Genetics and Genomics in Medicine (2nd Edition), CRC Press Taylor&Francis Group, Boca Raton London New York.
4. Thompson & Thompson Genetics and Genomics in Medicine (2023), Elsevier eBook on VitalSource, 9th Edition.
5. Gil de Bona A, Karaaslan H (2022) – Human Tooth and Developmental Dental Defects, IntechOpen.

RECOMMENDED BIBLIOGRAPHY

6. Cummings MR (2019) – Human Heredity: Principles and Issues, 12th Edition, Thomson Brooks Cole Publishing
7. Gorlin RJ, Cohen MM, HenneKam RCM (2010) – Syndromes of the Head and Neck, 5th Ed., Oxford Univ Press
8. Rusu C., Pânzaru M.(2014) – Medical Genetics Practical Lessons,Performantica or Dracopoli N.C et al. (2004) – Short Protocols in HUMAN GENETICS , Wiley
9. Jones KL (2021) – SMITH'S Recognizable Patterns of Human Malformation, 8th Ed., Elsevier Saunders
10. Lewis Ricki (2017) - Human Genetics, 12th Edition, McGraw Hill Higher Education

9. Corroborating the contents of the discipline with the expectations of epistemic community representatives, professional associations and employers in the fields representative for the program

- The study program covers essential aspects of genetics, acquainting students with terminology, concepts, principles, and specific research methods in genetics, preparing them both theoretically and practically for their future medical careers.
- The curriculum promotes the development of professional and transversal competencies required in medical practice, both in Romania and the EU. Additionally, the selected topics are relevant to the needs and requirements of the epistemic community.
- The thematic content of the practical work and the course is approached multidisciplinary, stimulating independent and creative thinking, critical analysis, and is the result of collaboration with faculty members from other universities, both national and international.

10. Evaluation

10.1 Evaluation			
Activity type	**Evaluation Criteria**	**Methods of evaluation**	**% out of final grade**
Lecture	**A. Knowledge for mark 5:** A low level of competence characterized by modest knowledge of the subject matter and an inadequate use of specific terminology; fundamental concepts are understood to a very limited extent and are expressed with difficulty; the provided answers lack clear syntheses or an appropriate approach to the topics. This performance corresponds to obtaining 30 correct answers out of the 60 questions on the grid.	**Continuous Assessment** - Control Papers (Multiple Choice Test) 2 written tests in the form of multiple-choice tests covering the course material and practical work completed in the first and second halves of the semester.	**20%**

	B. Additional knowledge for mark 10 A high level of competence characterized by deep and extensive knowledge of the subject matter, along with impeccable use of specific terminology; the student demonstrates a detailed and rigorous understanding of fundamental concepts and can express them with clarity and precision; the provided answers are accompanied by clear and appropriate syntheses, highlighting a complex and comprehensive approach to the topics. This performance corresponds to obtaining nearly a perfect number of correct answers out of the 60 questions on the grid.	**Summative Assessment** - Written Theoretical Exam (multiple-choice or essay-type)	**50%**
Laboratory Sessions	**A. Knowledge for mark 5:** A low level of performance characterized by evident confusion regarding fundamental concepts and specific terminology; modest abilities and skills in using laboratory techniques; errors in solving practical applications. **B. Additional knowledge for mark 10:** A high level of performance characterized by excellence in knowledge and the application of working methods; the ability to choose and justify complex methods based on suspected pathology; efficient teamwork and taking on leadership roles; excellence in handling equipment and conducting experiments; the ability to solve complex problems and interpret data in-depth; an understanding of professional ethics.	**Practical assessment:** Continuous oral evaluation through formative assessments, including surveys and teacher-student dialogues, as well as a structured questionnaire. The student's conduct in the laboratory is also assessed, focusing on aspects such as attendance, punctuality, appropriate attire, theoretical preparation relevant to the task at hand, practical skills, and creativity.	**10%**
		Final Practical Assessment: An oral examination to evaluate the student's ability to apply theoretical knowledge in practice and to assess their proficiency in the skills acquired.	**20%**

Minimum performance standards

Students should possess a solid understanding of:

- The laws and molecular mechanisms governing heredity and human variability, the genotype-phenotype relationship, and the role of genetic factors in the development of oro-dento-craniofacial anomalies and genetic diseases.
- The principles of genetic analysis and the application of genetic tests based on the underlying cause of the disease or anomaly under investigation.
- The integration of Genetics/Genomics knowledge into modern precision medicine, including predictive, personalised, preventive, and participatory approaches.

Students should also demonstrate proficiency in laboratory work, both independently and as part of a team.

Date: August 22, 2024

Chair of Genetics Division,
Prof. dr. Emilia Severin

Department director,
Prof. dr. Marina Imre

Date of the approval in Department Board: