**FACULTY OF STOMATOLOGY**

**STUDY PROGRAM 0911.1 STOMATOLOGY**

**CHAIR OF MOLECULAR BIOLOGY AND HUMAN GENETICS**

APPROVED
at the meeting of the Commission for Quality Assurance and Evaluation of the Curriculum in Dentistry
Minutes No. 2 of 30.01.2025
Chairmen, PhD, DMS, Associate professor,
Zanoaga Oleg

APPROVED
at the Council meeting of the Faculty of Stomatology
Minutes No. 04 of 04.02.2025
Dean of Faculty, PhD, Associate Professor,
Solomon Oleg

APPROVED
approved at the meeting of the chair of Molecular Biology and Human Genetics
Minutes No.10 of 13.01.2025
Head of chair Associate professor, PhD
Cemortan Igor

FACULTATEA DE STOMATOLOGIE

# SYLLABUS

DISCIPLINE **HUMAN GENETICS**

**Integrated studies**

Type of course: **Compulsory discipline**

Curriculum developed by the team of authors:

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# I. INTRODUCTION

- **General presentation of the discipline: place and role of the discipline in the formation of the specific competences of the professional / specialty training program**

  The Human Genetics course is, throughout the world, one of the basic components in the curriculum of medical schools and can form the main paradigm in the medical education of dentistry students. This course highlights the role of the genome vs. environmental factors in the formation, development and functioning of the human body. The genotype of a person is unique and unrepeatable, it is set at the moment of fertilization - the combination of maternal and paternal genes. Genetic individuality determines biological individuality - the phenotype - the set of specific traits of a body, produced by permanent interaction, in different proportions of heredity and environment. Changes in genetic material - mutations - can cause different pathological conditions with the involvement of one or more systems, which can explain the occurrence of genetic diseases or syndromes of the cranio-facial area and teeth. There are two important genetic phenomena considered: family aggregation and hereditary transmission of normal and pathological characters; different vulnerability of people to external aggression, variable manifestations of a disease in different people, different reaction to certain pharmacological preparations.

- **Mission of the curriculum (aim) in professional training**

The course is structured to demonstrate that genetics can be the core of modern medicine that studies biological development and possible variations of the entire life cycle; genetics is of major importance in calculating genetic risk and preventing human disease; high resolution molecular techniques used in human genetics are the primary tool for studying diseases in medical specialties. Human Genetics ensures the possibility of

• elucidation of pathogenic mechanisms of genetic diseases and genetic predisposition (cranio-facial area, teeth, cancers);
• genotypic diagnosis: pre-symptomatic or prenatal;
• genomic pharmacology - blocking the expression or replication of mutant genes;
• gene therapy - the introduction of normal genes into somatic cells of patients with mutant genes;
• individualized disease prophylaxis;
• changing the doctor-patient relationship - "There are no sick people, but only families of sick people".

- Language of the discipline: Romanian, Russian and English.

- Beneficiaries: students of the 1st year, Faculty of Stomatology.

# II. MANAGEMENT OF THE DISCIPLINE

| | | | |
|---|---|---|---|
| Code of discipline | | **F.02.O.016** | |
| Name of the discipline | | **Human Genetics** | |
| Person(s) in charge of the discipline | | **Associate professor Igor Cemortan** | |
| Year | **I** | Semester/Semesters | **2** |
| Total number of hours, including: | | | **90** |
| Lectures | **15** | Practical/laboratory hours | **15** |
| Seminars | **15** | Self-training | **45** |
| Form of assessment | **E** | Number of credits | **3** |

# III. TRAINING AIMS WITHIN THE DISCIPLINE

***At the end of the discipline study the student will be able to:***

***at the level of knowledge and understanding:***

- have knowledge of the processes of storing, transmitting and expressing genetic information through which the morpho-functional traits specific to each individual are realized;
- know the molecular and cellular substrate of heredity - DNA and chromosomes - that contain, express and transmit hereditary information;
- understand the transmission of hereditary information in the succession of generations of cells and organisms;
- understand phenomena that cause genetic differences between individuals of a population and between different populations: mutations, genetic recombination in meiosis and fertilization, migration - the flow of genes in the receiving population;
- know the nomenclature of mutations;
- know of the heredity laws for normal and pathological monogenic, polygenic and multifactorial traits;
- know the principles of different techniques used in human genetics.

***at the application level:***

- determine normal vs abnormal karyotype;
- establish the nature of aneuploidy: mitotic error or meiotic error;
- determine the type of mutation, possible consequences, identification methods;
- evaluate the genetic phenomena involved in the phenotypic manifestation of normal and pathological genes: gene interactions, pleiotropy, penetrance, expressivity, heterogeneity, imprinting, anticipation.
- know the principles and practical applications of the karyotyping technique;
- evaluate the Barr body test;
- To apply the laws of inheritance of normal monogenically determined polymorphic traits:
  - blood groups (variations of erythrocyte Ag);
  - serum groups (variations of plasma proteins);
  - enzyme groups (isoenzymatic variations of an enzyme);
  - tissue groups (HLA).;
- To analyze the genealogical inheritance of normal or pathological traits, to determine the type of inheritance, the genotype and to calculate the risk of recurrence of a monogenic disease;
- To evaluate the results of different molecular-genetic techniques: PCR, Southern-blot and DNA sequencing;
- To apply the methods of studying hereditary traits in different case studies:
  - a twin method;
  - statistical population method.

- ***at the integration level:***

- Understand the theoretical importance of studying hereditary characters:
  - different vulnerability to external aggression;
  - genetic predisposition to certain diseases;
  - variable manifestations of a disease;
  - Different treatment response.

- Understand the practical importance of studying hereditary characters:
  - genetic approach to the patient in various clinical disciplines: cardiology, neurology, pediatrics, nephrology, pulmonology, psychiatry, oncology, etc.
  - prevention of hemolytic disease of the newborn caused by Rh system incompatibility;
  - paternity and parentage tests;
  - people identification;
  - diagnosis of the type of twins.

## IV. PROVISIONAL TERMS AND CONDITIONS

Requirements for first year, second semester students:

- knowledge of the language of studies;
- confirmed competences in sciences (biology, chemistry, physics) at the school level;
- competences in molecular biology;
- competences in histology and anatomy;
- digital competences (use of the Internet, document processing, electronic tables and presentations, use of graphics programs);
- ability to communicate and work in a team;
- qualities - tolerance, compassion, autonomy.

# V. THEMES AND ESTIMATEALLOCATION OF HOURS

***Lectures, practical hours/seminars and self-training***

| No. d/o | THEME | Number of hours | | |
|---|---|---|---|---|
| | | Lectures | Seminars/ Practical hours | Self-training |
| 1. | Human genetics and its importance in stomatology. Human, heredity and environment. The genetic and biological uniqueness of every human being. Genetic apparatus of human cell. Nuclear and mitochondrial genome. | 1 | 2 | 3 |
| 2. | Morphology and structure of human chromosomes. Chromosomal nomenclature. Sexual chromosomes. Normal human karyotype. | 1 | 2 | 3 |
| 3. | Methods of studying of human chromosomes. Evaluation of the normal and pathological human karyotypes. | 1 | 2 | 3 |
| 4. | Numeric and structural chromosome abnormalities. | 1 | 2 | 3 |
| 5. | Dynamics of chromosomes during mitosis. Errors of distribution of genetic information during mitosis. Chromosomal mosaics. | 1 | 2 | 3 |
| 6. | Genetic events during meiosis. Transferring of genetic information from ascendant to offspring. Errors of meiosis and their consequences. | 1 | 2 | 3 |
| 7. | Human genes: structure, functions and location. Genes responsible for development and function on cranio-facial area and teeth. | 1 | 2 | 3 |
| 8. | Methods of direct and indirect gene analysis. | 1 | 2 | 3 |
| 9. | Gene mutations. Classification of mutations. Point mutations. Phenotypic consequences of gene mutations in cranio-facial area and teeth. | 1 | 2 | 3 |
| 10. | Gene functions and relation genotype-phenotype. Pleiotropic and polygenic characters. Genetic heterogeneity (allelic and locus). Gene's interactions: allelic, non-allelic and interaction between genes and environment. | 1 | 2 | 3 |
| 11. | Normal monogenic human traits. Inheritance of the systems: AB0, Rh, MNS, Hp, Xg, HLA. Polygenic inheritance. | 1 | 2 | 3 |
| 12. | Study of human hereditary traits. Hardy-Weinberg low. Twins studies. Applications of the populational-statistic method and twins' studies. | 1 | 2 | 3 |
| 13. | Human pathological monogenic traits which affect cranio-facial region and teeth. Types of inheritance. Criteria for the recognition of dominant and recessive, autosomal and X-linked inheritance. | 1 | 2 | 3 |
| 14. | Inheritance of human pathological traits with incomplete penetrance, variable expression, genetic heterogeneity. | 1 | 2 | 3 |
| 15. | Genetic counselling. Prenatal diagnosis. | 1 | 2 | 3 |

| No. d/o | THEME | Number of hours | | |
|---|---|---|---|---|
| | | Lectures | Seminars/ Practical hours | Self-training |
| **Total** | | **15** | **30** | **45** |

## VI. REFERENCE OBJECTIVES OF CONTENT UNITS

| Objectives | Content units |
|---|---|
| **Theme (chapter) 1. Normal and pathological human karyotype** | |
| • To define genome, genotype, plasmotype, phenotype;<br>• to know the normal karyotype and the chromosomal polymorphisms;<br>• to know the nomenclature and consequences of numerical and structural chromosomal abnormalities;<br>• to demonstrate the difference between autosomal and gonosomal abnormalities, balanced and unbalanced abnormalities;<br>• to know the dynamics of chromosomes in mitosis and meiosis, errors and their cytogenetic consequences;<br>• to comment on the medical significance of knowing the mitotic or meiotic origin of the chromosomal abnormality;<br>• to know the principles and stages of different cytogenetic techniques;<br>• to interpret the karyogram, the Barr body test, the F body test, the FISH test;<br>• to apply knowledge to other disciplines;<br>• to formulate conclusions;<br>• to develop own opinions | Genetic apparatus of human cell. Nuclear and mitochondrial genome.<br>Human chromosomes, structure and nomenclature. Sexual chromosomes. Normal human karyotype.<br>Methods of studying of human chromosomes. Numeric and structural chromosomal abnormalities.<br>Dynamics of chromosomes during mitosis. Errors of distribution of genetic information during mitosis. Chromosomal mosaics.<br>Transferring of genetic information from ascendant to offspring. Genetic events during meiosis. Errors of recombination and distribution of chromosomes during meiosis.<br>Evaluation of the normal and pathological human karyotypes |
| **Theme (chapter) 2. Peculiarities of human genes** | |
| • To define gene, gene expression, genotype, homozygote, heterozygote, hemizigot, phenotype<br>• to know the particularities of organization, properties and functions of different genes;<br>• to know and understand the causes, mechanisms and nomenclature of gene mutations; | Human genes: structure, functions and location. Methods of direct and indirect gene analysis. Gene mutations. Point mutations. Phenotypic consequences of gene mutations.<br>Normal monogenic human traits (I). Human blood types (AB0, Rh).<br>Normal monogenic human traits (II). Inheritance of the systems: MNS, Hp, Xg, HLA. |

| Objectives | Content units |
|---|---|
| • to know the consequences of mutations of coding, regulatory and modulating regions of human genes;<br>• to demonstrate the dominant, codominant or recessive effect of molecular mutations at the molecular, cellular and organism levels.<br>• to understand the principles, stages and components needed for different gene analysis techniques;<br>• to model PCR technique, Southern blot technique, dideoxy sequencing;<br>• to interpret the results of gene analysis in various techniques;<br>• to apply the knowledge gained to other disciplines | Polygenic inheritance. Dermatoglyphs and their applications. |
| **Theme (chapter) 3. Study of hereditary traits** | |
| • To define gene pool, MZT concordance, DZT concordance, AD, AR, XD, XR gene transmission;<br>• To now the methods of confirming the genetic nature of a normal or pathological trait;<br>• To build and analyze genealogical trees with the study of heredity of different phenotypes;<br>• To understand the genetic phenomena that may occur in the manifestation and transmission of traits: incomplete penetrance, variable expression, allelic / non-allelic heterogeneity, anticipation;<br>• to apply the population-statistical method in calculating the frequency of pathological genes and carriers in the RM population;<br>• to apply the twin method in calculating the share of genetic and environmental factors in the manifestation of normal or pathological traits;<br>• to apply the genealogical method in determining the type of transmission and calculating the risk of recurrence of monogenic, Mendelian diseases;<br>• to understand the role and place of genetic counseling in addressing the patient, the couple, healthy people;<br>• to understand the indications and limits of prenatal diagnosis. | Study of human hereditary traits. Hardy-Weinberg low. Twins studies. Applications of the populational-statistic method and twins studies.<br>Inheritance of pathological monogenic traits. Criteria for the recognition of dominant and recessive, autosomal and X-linked inheritance.<br>Inheritance of human pathological traits with incomplete penetrance, variable expression, genetic heterogeneity.<br>Genetic counselling. Prenatal diagnosis. |

# VII. PROFESSIONAL (SPECIFIC (SC)) AND TRANSVERSAL (TC) COMPETENCES AND STUDY FINALITIES

## ✓ Professional (specific) (CP) competences

**CP1. The responsible execution of professional tasks with the application of the values and norms of professional ethics, as well as the provisions of the legislation in force.** Apply the legal and normative framework in practical activity. Respects the norms of ethics and deontology. It ensures compliance with ethical and deontological norms and is guided by the provisions of the code of medical ethics. Promote collegial relationships with co-workers. Carry out free and independent activities according to the oath of the medical profession. Knows and respects the rights and technical rules regarding the sanitary-hygienic and anti-epidemic regime in various socio-medical situations according to the legislation in force. Knows and respects the provisions of the collective labor agreement, the protection rules and the safety and health technique at the workplace. It ensures the compliance and correctness of the fulfillment of service obligations in the provision of care to the population in public, private and community medical and sanitary institutions. It encourages informed ethical decision making and respects the patient's decision.

**CP2. Adequate knowledge of the sciences about the structure of the body, the physiological functions and the behavior of the human body in various physiological and pathological states, as well as the existing relationships between the state of health, the physical and the social environment.** Knows the structures, physiological functions of organs and organ systems in healthy subjects. Recognizes the physiological and pathological processes of the human being and the psychosocial responses of individuals in various states of health. Knows the relevant terminology for the important signs and symptoms that are derived from various pathophysiological conditions. Identifies pathophysiological processes and their expression, as well as risk factors that determine health and disease at different stages of the life cycle. Appreciate the relationship between the state of health, the physical and social environment of the human being. Knows the possible evolution and complications leading to the main pathological processes.

**CP6. Conducting scientific research in the field of health and other branches of science.** Plans, organizes and executes scientific research in the field. Identifies sources of information, selects research materials and methods, performs experiments, statistical processing of research results, formulation of conclusions and proposals. Elaborates and supports speeches, presentations at scientific events by demonstrating personal attitude, coherence in exposition and scientific correctness; participates in discussions and debates at scientific events.

**CP7. Promoting and ensuring the prestige of the medical profession and raising the professional level.** Plans, organizes and executes scientific research in the field. Identifies sources of information, selects research materials and methods, performs experiments, statistical processing of research results, formulation of conclusions and proposals. Elaborates and supports speeches, presentations at scientific events by demonstrating personal attitude, coherence in exposition and scientific correctness; participates in discussions and debates at scientific events. It achieves the maintenance of the high level of professional skills during the entire period of activity. Actively participates in professional associations for the purpose of correctly fulfilling professional obligations, promoting the image of the doctor and the medical system in society. It contributes to the adjustment of the legislative framework in the field of medical assistance to European standards, ensuring the quality of the medical act, implementing the Rules of Good Practice, promoting the image of the medical profession at scientific-practical events and in the mass media.

## ✓ Transversal competences (CT)

**CT1. Autonomy and responsibility in activity.** The application of rigorous and efficient work rules, the manifestation of a responsible attitude towards the performance of professional tasks with the application of the values and norms of professional ethics, as well as the provisions of the legislation in force. Promoting logical reasoning, practical applicability, evaluation and self-evaluation in decision-making.

## ✓ Study finalities

- to determine normal and abnormal karyotype;
- to determine the nature of the chromosomal anomaly - mitotic error, meiotic error;
- to know the principles and practical applications of cytogenetic techniques;
- to apply the heredity laws for normal and pathological monogenic characters;
- to know the nomenclature of chromosomal and gene mutations;
- to analyze the genealogical transmission of normal or pathological traits and determine the type of inheritance, genotype and calculate the risk of recurrence of a genetic disease;
- to apply the methods studied in determining the genetic nature of different traits.

## VIII. STUDENT'S SELF-TRAINING

| No. | Expected product | Implementation strategies | Assessment criteria | Implementation terms |
|---|---|---|---|---|
| 1 | Working with information sources: | Reading the lecture or the material from the textbook on the topic carefully.<br>Reading questions on the topic, which require a reflection on the subject.<br>To get acquainted with the list of additional information sources on the topic. Select the source of additional information for the topic.<br>Reading the text entirely, carefully and writing the main content.<br>Writing generalizations and conclusions regarding the importance of the topic / subject. | Ability to extract the main information; interpretative skills; the volume of work | During the semester |
| 2 | Working with the Workbook: | Before solving the tasks in the workbook to analyze the information and images from the respective subject in the lecture and textbook. Solving consecutive tasks. Formulate conclusions at the end of each lesson. Verifying the final conclusions of the lesson and appreciating their fulfilment. Selection of additional information, using electronic addresses and additional bibliography. | Workload, problem solving, ability to formulate conclusions | During the semester |
| 3 | Working with online materials | Online self-assessment, study of online materials on the Chair site, expressing own opinions on forum and chat | Number and duration of chair site entries, self-evaluation results | During the semester |

# IX. METHODOLOGICAL SUGGESTIONS FOR TEACHING-LEARNING-ASSESSMENT

- ***Teaching methods used***

In teaching Human Genetics are used various didactic methods and techniques, oriented towards the efficient learning and achievement of the objectives of the didactic process. During theoretical lectures, along with traditional methods (lecture-exposure, lecture-conversation, synthesis lecture), modern methods (lesson-debate, lecture-conference, problem-lesson) are also used. Forms of individual, frontal, group, virtual lab work are used in the practical classes. To learn the material in depth, different semiotic systems (scientific language, graphical and computerized language) and teaching materials (tables, diagrams, micro-pictures, transparent films) are used. During the lessons and extracurricular activities are used Communication Information Technologies - PowerPoint presentations, on-line lessons.

## *Learning methods used*

- **Observation** - Identification of elements characteristic to some structures or biological phenomena, describing these elements or phenomena.
- **Analysis** - Imaginary decomposition of the whole into component parts. Highlighting the essential elements. Studying each element as part of the whole.
- **Diagram / picture analysis** - Selection of required information. Recognition based on knowledge and selected information of the structures indicated in the diagram, drawing. Analysis of the functions / role of recognized structures.
- **Comparison** - Analysis of the first object / process in a group and determining its main features. Analysis of the second object / process and determining its main features. Comparing objects / processes and highlighting common features. Comparing objects / processes and determining differences. Establishing distinguishing criteria. Formulating conclusions.
- **Classification** - Identification of the structures / processes to be classified. Determining the criteria on which classification is to be made. Distribution of structures / processes by groups according to established criteria.
- **Scheme drawing** - Selection of elements, which must be included in the scheme. Showing the selected elements through different symbols / colors and showing their relationships. Formulating an appropriate title and legend for the symbols used.
- **Modeling** - Identifying and selecting the elements needed for modeling the phenomenon. Imaging (graphically, schematically) the phenomenon studied. Realizing the phenomenon using the developed model. Formulating conclusions, deduced from arguments or findings.
- **Experiment** - Formulating a hypothesis, based on known facts, on the process / phenomenon studied. Verifying the hypothesis by performing the processes / phenomena studied under laboratory conditions. Formulation of conclusions deduced from arguments or findings.

- ***Applied teaching strategies / technologies*** *(specific to the discipline)*

- "Brainstorming", "Multi-voting"; "The round table"; "Group Interview"; "Case Study"; "Creative Controversy"; "Focus-group technique", "Portfolio".
- Virtual Practices

- ***Methods of assessment*** *(including the method of final mark calculation)*

✓ **Current**: frontal and / or individual control through

- (a) applying docimological tests,

- (b) solving problems / exercises,
- (c) analysis of case studies
- (d) performing role-plays on the topics discussed.
- (e) tests
- (e) paper tests

***Final**:* exam.

The **final mark** will consist of the average mark of three concluding tests (50%), and the final test in computerized system (50%).

The average mark and the marks of all the final exam stages (computer, written test) - will be expressed in numbers according to the marks scale (as in the table) and the final mark obtained will be expressed in two decimals and will be written in the marks book.

**Method of mark rounding at different assessment stages**

| Intermediate marks scale (annual average, marks from the examination stages) | National Assessment System | ECTS Equivalent |
|---|---|---|
| **1,00-3,00** | **2** | **F** |
| **3,01-4,99** | **4** | **FX** |
| **5,00** | **5** | **E** |
| **5,01-5,50** | **5,5** | |
| **5,51-6,0** | **6** | |
| **6,01-6,50** | **6,5** | **D** |
| **6,51-7,00** | **7** | |
| **7,01-7,50** | **7,5** | **C** |
| **7,51-8,00** | **8** | |
| **8,01-8,50** | **8,5** | **B** |
| **8,51-9,00** | **9** | |
| **9,01-9,50** | **9,5** | **A** |
| **9,51-10,0** | **10** | |

*Absence on examination without good reason is recorded as "absent" and is equivalent to 0 (zero). The student has the right to have two re-examinations.*

# X. RECOMMENDED LITERATURE:

## *A. Compulsory:*

1. Human Genetics. Exercise book Capcelea S., Perciuleac L., Cemortan I, 2024
2. Presentations of lectures: SIMU
3. Reading materials: www.biologiemoleculară.usmf.md
4. On line-tests: www.e.usmf. md

## *B. Additional*

1. Jameson J. L., Fauci A. S., Kasper D. L., Hauser S. L., Longo D. L., Loscalzo J. Harrison's principles of internal medicine. 20e. PART 16 Genes, the Environment, and Disease, 2019
2. https://ghr.nlm.nih.gov/
3. http://www.genecards.org/
4. https://www.malacards.org/
5. https://www.ncbi.nlm.nih.gov/pubmed/
6. https://genome.cshlp.org/
7. Genes. B. Lewin, 2017
8. Genetics (4 volumes), Robinson R., 2003
9. Human Molecular Genetics, Strachan T., Read A., 2001