



**CD 8.5.1 CURRICULUM DISCIPLINĂ PENTRU  
STUDII UNIVERSITARE**

**Redacția: 08**

**Data: 21.02.2020**

**Pag. 1/14**

**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 Faculty of Stomatology  
Minutes No. \_\_\_ of \_\_\_\_\_

Committee president, PhD, Associate professor,  
Stepco Elena \_\_\_\_\_

**APPROVED**

at the Council meeting of the Faculty of Stomatology  
Minutes No. \_\_\_ of \_\_\_\_\_

Dean of Faculty, PhD, Associate professor,  
Solomon Oleg \_\_\_\_\_

**APPROVED**

approved at the meeting of the chair of Molecular Biology and Human Genetics  
Minutes No.1 of 24.08.2020

Head of chair Associate professor, PhD  
Cemortan Igor \_\_\_\_\_

**CURRICULUM**

**DISCIPLINE HUMAN GENETICS**

**Integrated studies**

Type of course: **Compulsory discipline**

Chisinau, 2019



## 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 course: Romanian, Russian and English.
  - Beneficiaries: students of the 1<sup>st</sup> year, Faculty of Stomatology.



**CD 8.5.1 CURRICULUM DISCIPLINĂ PENTRU  
STUDII UNIVERSITARE**

**Redacția: 08**

**Data: 21.02.2020**

**Pag. 3/14**

**II. MANAGEMENT OF THE DISCIPLINE**

Code of discipline		<b>F.02.O.017</b>	
Name of the discipline		<b>Human Genetics</b>	
Person(s) in charge of the discipline		<b>Associate professor Igor Cemortan</b>	
Year	<b>I</b>	Semester/Semesters	<b>2</b>
Total number of hours, including:			<b>90</b>
Lectures	<b>17</b>	Practical/laboratory hours	<b>17</b>
Seminars	<b>17</b>	Self-training	<b>39</b>
Clinical internship			<b>-</b>
Form of assessment	<b>E</b>	Number of credits	<b>3</b>



### 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;
- understand the genetic basis of human body development;
- understand the genetic basis of the immune response;
- know the genetic mechanisms of cancerogenesis;
- 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;

*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.



## CD 8.5.1 CURRICULUM DISCIPLINĂ PENTRU STUDII UNIVERSITARE

Redacția: 08

Data: 21.02.2020

Pag. 5/14

### 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.



**CD 8.5.1 CURRICULUM DISCIPLINĂ PENTRU  
STUDII UNIVERSITARE**

**Redacția: 08**

**Data: 21.02.2020**

**Pag. 6/14**

**V. THEMES AND ESTIMATE ALLOCATION OF HOURS**

**A. Lectures**

	<b>Topic</b>	<b>Ore</b>
1.	Human genetics and its importance in stomatology. Human, heredity and environment.	1
2.	Morphology and structure of human chromosomes. Methods of the human chromosome analysis. Identification, classification and nomenclature of human chromosomes. Sexual chromosomes.	2
3.	Chromosome abnormalities. Abnormalities in chromosome number. Abnormalities in chromosome structure. Chromosomal syndromes which affect cranio-facial area and teeth.	2
4.	Inheritance of genetic information from cell to cell. Segregation of chromosomes during mitosis. Errors of distribution of genetic information during mitosis. Chromosomal mosaics.	1
5.	Transferring of genetic information from ascendant to offspring. Genetic events during meiosis. Errors of recombination and distribution of chromosomes during meiosis. Particularities of meiosis in men and women. Errors during fecundation.	1
6.	Human genes: structure, functions and location. Locus. Allelic genes. Polyallelism. Gene linkage.	2
7.	Molecular analysis of human genes	2
8.	Gene mutations. Phenotypic consequences of gene mutations.	2
9.	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.	2
10.	Inheritance of monogenic characters. Sex-linked inheritance. Non-mendelian transmission. Mitochondrial inheritance. Genomic imprinting. Uniparental di-somy.	1
11.	General information about genetic pathology. Genetic diseases which affect cranio-facial area and teeth. Classification and frequency of diseases. General characterization of genetic diseases. Methods of studying and diagnostic of genetic diseases. Genetic counselling. Prevention of the genetic diseases. Prenatal diagnostic.	1
	<b>Total</b>	<b>17</b>

**B. Practical hours/seminars and self-training**

No. d/o	THEME	Number of hours		
		Seminars	Practical hours	Self-training
1.	Genetic apparatus of human cell. Nuclear and mitochondrial genome.	1	1	3
2.	Morphology and structure of human chromosomes. Chromosomal nomenclature. Sexual chromosomes. Normal human karyotype.	1	1	3
3.	Methods of studying of human chromosomes.	1	1	3
4.	Numeric and structural chromosome abnormalities.	1	1	2
5.	Dynamics of chromosomes during mitosis. Errors of distribution of genetic information during mitosis. Chromosomal mosaics.	1	1	2
6.	Genetic events during meiosis. Transferring of genetic information from ascendant to offspring. Errors of meiosis and their consequences.	1	1	2



**CD 8.5.1 CURRICULUM DISCIPLINĂ PENTRU  
STUDII UNIVERSITARE**

**Redacția: 08**

**Data: 21.02.2020**

**Pag. 7/14**

No. d/o	THEME	Number of hours		
		Seminars	Practical hours	Self- training
7.	Evaluation of the normal and pathological human karyotypes	1	1	3
8.	Human genes: structure, functions and location. Genes responsible for development and function on cranio-facial area and teeth.	1	1	2
9.	Methods of direct and indirect gene analysis.	1	1	3
10.	Gene mutations. Classification of mutations. Point mutations. Phenotypic consequences of gene mutations in cranio-facial area and teeth.	1	1	2
11.	Normal monogenic human traits (I). Human blood types (ABO, Rh).	1	1	2
12.	Normal monogenic human traits (II). Inheritance of the systems: MNS, Hp, Xg, HLA.	1	1	2
13.	Polygenic inheritance.	1	1	2
14.	Study of human hereditary traits. Hardy-Weinberg law. Twins studies. Applications of the populational-statistic method and twins' studies.	1	1	2
15.	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	1	2
16.	Inheritance of human pathological traits with incomplete penetrance, variable expression, genetic heterogeneity.	1	1	2
17.	Genetic counselling. Prenatal diagnosis.	1	1	2
<b>Total</b>		<b>17</b>	<b>17</b>	<b>39</b>



## VI. REFERENCE OBJECTIVES OF CONTENT UNITS

Objectives	Content units
<b>Theme (chapter) 1. Normal and pathological human karyotype</b>	
<ul style="list-style-type: none"><li>• To define genome, genotype, plasmotype, phenotype;</li><li>• to know the normal karyotype and the chromosomal polymorphisms;</li><li>• to know the nomenclature and consequences of numerical and structural chromosomal abnormalities;</li><li>• to demonstrate the difference between autosomal and gonosomal abnormalities, balanced and unbalanced abnormalities;</li><li>• to know the dynamics of chromosomes in mitosis and meiosis, errors and their cytogenetic consequences;</li><li>• to comment on the medical significance of knowing the mitotic or meiotic origin of the chromosomal abnormality;</li><li>• to know the principles and stages of different cytogenetic techniques;</li><li>• to interpret the karyogram, the Barr body test, the F body test, the FISH test;</li><li>• to apply knowledge to other disciplines;</li><li>• to formulate conclusions;</li><li>• to develop own opinions</li></ul>	<ol style="list-style-type: none"><li>1. Genetic apparatus of human cell. Nuclear and mitochondrial genome.</li><li>2. Human chromosomes, structure and nomenclature. Sexual chromosomes. Normal human karyotype.</li><li>3. Methods of studying of human chromosomes.</li><li>4. Numeric and structural chromosomal abnormalities.</li><li>5. Dynamics of chromosomes during mitosis. Errors of distribution of genetic information during mitosis. Chromosomal mosaics.</li><li>6. Transferring of genetic information from ascendant to offspring. Genetic events during meiosis. Errors of recombination and distribution of chromosomes during meiosis.</li><li>7. Evaluation of the normal and pathological human karyotypes</li></ol>
<b>Theme (chapter) 2. Human genes</b>	
<ul style="list-style-type: none"><li>• To define gene, gene expression, genotype, homozygote, heterozygote, hemizigot, phenotype</li><li>• to know the particularities of organization, properties and functions of different genes;</li><li>• to know and understand the causes, mechanisms and nomenclature of gene mutations;</li><li>• to know the consequences of mutations of coding, regulatory and modulating regions of human genes;</li><li>• to demonstrate the dominant, codominant or recessive effect of molecular mutations at the molecular, cellular and organism levels.</li><li>• to understand the principles, stages and components needed for different gene analysis techniques;</li><li>• to model PCR technique, Southern blot technique, dideoxy sequencing;</li></ul>	<ol style="list-style-type: none"><li>1. Human genes: structure, functions and location.</li><li>2. Methods of direct and indirect gene analysis.</li><li>3. Gene mutations. Point mutations. Phenotypic consequences of gene mutations.</li><li>4. Normal monogenic human traits (I). Human blood types (AB0, Rh).</li><li>5. Normal monogenic human traits (II). Inheritance of the systems: MNS, Hp, Xg, HLA.</li><li>6. Polygenic inheritance. Dermatoglyphs and their applications.</li></ol>





**CD 8.5.1 CURRICULUM DISCIPLINĂ PENTRU  
STUDII UNIVERSITARE**

**Redacția: 08**

**Data: 21.02.2020**

**Pag. 9/14**

**Objectives**

**Content units**

- to interpret the results of gene analysis in various techniques;
- to apply the knowledge gained to other disciplines

**Theme (chapter) 3. Study of the human hereditary traits**

- To define gene pool, MZT concordance, DZT concordance, AD, AR, XD, XR gene transmission;
- To know the methods of confirming the genetic nature of a normal or pathological trait;
- To build and analyze genealogical trees with the study of heredity of different phenotypes;
- To understand the genetic phenomena that may occur in the manifestation and transmission of traits: incomplete penetrance, variable expression, allelic / non-allelic heterogeneity, anticipation;
- to apply the population-statistical method in calculating the frequency of pathological genes and carriers in the RM population;
- to apply the twin method in calculating the share of genetic and environmental factors in the manifestation of normal or pathological traits;
- to apply the genealogical method in determining the type of transmission and calculating the risk of recurrence of monogenic, Mendelian diseases;
- to understand the role and place of genetic counseling in addressing the patient, the couple, healthy people;
- to understand the indications and limits of prenatal diagnosis.

1. Study of human hereditary traits. Hardy-Weinberg law. Twins studies. Applications of the populational-statistic method and twins studies.
2. Human pathological monogenic traits. Types of inheritance. Criteria for the recognition of dominant and recessive, autosomal and X-linked inheritance.
3. Inheritance of human pathological traits with incomplete penetrance, variable expression, genetic heterogeneity.
4. Genetic counselling. Prenatal diagnosis.



**CD 8.5.1 CURRICULUM DISCIPLINĂ PENTRU  
STUDII UNIVERSITARE**

**Redacția: 08**

**Data: 21.02.2020**

**Pag. 10/14**

**VII. PROFESSIONAL (SPECIFIC (SC)) AND TRANSVERSAL (TC)  
COMPETENCES AND STUDY OUTCOMES**

✓ **Professional (specific) (SC) competences**

SC 1 Knowledge and understanding of the organization of the human genome, chromosomes and genes;

SC 2. Knowledge of human karyotype and principles of cytogenetic techniques. Explanation and interpretation of karyograms.

SC 3. Knowledge of the gene and chromosomal mutation nomenclature. Analysis and interpretation of various genetic tests.

SC 5. Comparing the results obtained by different cytogenetic and molecular techniques.

✓ **Study outcomes**

- 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 know the indications and limits of the PCR, Southern blot and Sanger techniques;
- 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.



**CD 8.5.1 CURRICULUM DISCIPLINĂ PENTRU  
STUDII UNIVERSITARE**

**Redacția: 08**

**Data: 21.02.2020**

**Pag. 11/14**

**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. Reading questions on the topic, which require a reflection on the subject. To get acquainted with the list of additional information sources on the topic. Select the source of additional information for the topic. Reading the text entirely, carefully and writing the main content. 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,



**CD 8.5.1 CURRICULUM DISCIPLINĂ PENTRU  
STUDII UNIVERSITARE**

**Redacția: 08**

**Data: 21.02.2020**

**Pag. 13/14**

- (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
<b>1,00-3,00</b>	<b>2</b>	<b>F</b>
<b>3,01-4,99</b>	<b>4</b>	<b>FX</b>
<b>5,00</b>	<b>5</b>	<b>E</b>
<b>5,01-5,50</b>	<b>5,5</b>	
<b>5,51-6,0</b>	<b>6</b>	
<b>6,01-6,50</b>	<b>6,5</b>	<b>D</b>
<b>6,51-7,00</b>	<b>7</b>	
<b>7,01-7,50</b>	<b>7,5</b>	<b>C</b>
<b>7,51-8,00</b>	<b>8</b>	
<b>8,01-8,50</b>	<b>8,5</b>	<b>B</b>
<b>8,51-8,00</b>	<b>9</b>	
<b>9,01-9,50</b>	<b>9,5</b>	<b>A</b>
<b>9,51-10,0</b>	<b>10</b>	

The average annual mark and the marks of all stages of final examination (computer assisted, test, oral) - are expressed in numbers according to the mark scale (according to the table), and the final mark obtained is expressed in number with two decimals, which is transferred to student's record-book.

*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.*



**CD 8.5.1 CURRICULUM DISCIPLINĂ PENTRU  
STUDII UNIVERSITARE**

**Redacția: 08**

**Data: 21.02.2020**

**Pag. 14/14**

**X. RECOMMENDED LITERATURE:**

*A. Compulsory:*

1. Human Genetics. Exercise book Capcelea S., Perciuleac L., Cemortan I, 2019
2. Presentations of lectures: [www.biologiemoleculara.usmf.md](http://www.biologiemoleculara.usmf.md)
3. Reading materials: [www.biologiemoleculara.usmf.md](http://www.biologiemoleculara.usmf.md)
4. On line-tests: [www.e.usmf.md](http://www.e.usmf.md)
5. <https://ghr.nlm.nih.gov/>

*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. <http://www.genecards.org/>
3. <https://www.malacards.org/>
4. <https://www.ncbi.nlm.nih.gov/pubmed/>
5. <https://genome.cshlp.org/>
6. Genes. B. Lewin, 2017
7. Genetics (4 volumes), Robinson R., 2003
8. Human Molecular Genetics, Strachan T., Read A., 2001