

Subject outline

1. Identification data of the subject

1.1 Higher education institution	"Ion Ionescu de la Brad" University of Life Sciences of Iasi
1.2 Faculty	Veterinary Medicine
1.3 Department	Preclinics
1.4 Field	Veterinary Medicine
1.5 Cycle of studies	Bachelor studies
1.6 Studies programme	Veterinary Medicine

2. Information regarding the subject

2.1 Name	Medical Genetics and Eredopathology						
2.2 Lecture coordinator	<i>Associate Professor Andrei Cristian GRĂDINARU</i>						
2.3 Practical activities coordinator	<i>Associate Professor Andrei Cristian GRĂDINARU</i>						
2.4 Year of study	II	2.5 Semester	IV	2.6 Evaluation type	Sumative	2.7 Subject status	Compulsory

3. Structure of the subject (hours/semester of didactic activities)

3.1 Number of hours/week	4	out of which:	2	3.3 <i>Seminars/practical works</i>	2
		3.2 Lecture			
3.4 Total number of hours in curricula	56	out of which:	28	3.6 <i>Seminars/practical works</i>	28
		3.5 Lecture			
Time distribution					hours
Study by manual, bibliography and lecture notes					26
Additional study in library, specialty electronic sources and on the field activities					10
Preparing for laboratory activities, homework, projects, portfolios and essays					10
Tutoring					2
Examinations					3
Other activities					5
3.7 Total hours of individual study	60				
3.9 Total hours/semester	56				
3.10 Number of credits	3				

4. Pre-requisites

4.1 Curriculum	• <i>Cell Biology, Biochemistry, Molecular Biology, Math</i>
4.2 Competencies	• <i>Molecular Biology</i>

5. Conditions

5.1. For conducting the lectures	The use of mobile phones and laptops is not allowed during the course lecture. It is not allowed to be late to the course, as well as leaving it without good reasons.
5.2. For conducting the practical activities	Students must wear a protective white coat. All practical applications will be led and supervised by the practical activity coordinator. The use of mobile phones is only allowed on the computer function, when requested by the practical activities coordinator. Arriving late or leaving the classroom without a valid reason is not permitted.

6. Acquired specific competences

Professional competencies	<ul style="list-style-type: none"> Gaining the information about <i>Fundamental Cytogenetics</i>, including cell' structure, cellular organelles with genetic functions, the nuclear and cytoplasmic genetic material and its pathways of expression and inheritance, of elements of classical and modern cytogenetic diagnosis. Analysis and interpretation of segregation schemes of various normal and pathological characters and traits. Investigations on the Genetic structure of various populations for different characters and traits, including Hardy-Weinberg equilibrium testing and frequencies of genes and genotypes. Diagnosis of numerical and structural chromosomal abnormalities and their phenotype expressing. The analysis of medical pedigree and the estimation of genetic risk for pathological traits inheritance.
Cross competencies	<ul style="list-style-type: none"> Gaining skills to work with specialized knowledge in <i>Genetics</i> field and to associate them with those in related subjects, such as <i>Cell Biology, Molecular Biology, Biochemistry, and Pathology</i>. Developing interest for animal phenotype interpretation, based both on genotype structure, and genotype and environment interaction.

7. Subject objectives

7.1 General objective of the subject	Acquiring of theoretical and practical knowledge in the field of Medical Veterinary Genetics, including elements of Cytogenetics, Molecular Genetics, Populational Genetics, Quantitative Genetics and Eredopathology.
7.2 Specific objectives	<p>Explaining and interpreting the results of segregation schemes.</p> <p>Establishing the genetic structure of animal populations.</p> <p>Evaluating the inheritance of various traits and characters.</p>

8. Content

Crt. no.	8.1 Lectures	Teaching methods	No. of hours
1.	<p>Chapter 1: THE CONTENT OF THE SUBJECT OF MEDICAL GENETICS AND EREDOPATHOLOGY</p> <p>1.1. The Science of Genetics: definition, object of study, heredity and variability</p> <p>1.2. Historical milestones in the evolution of the science of Genetics</p>	Power Point presentation, oral presentation	2
2.	<p>Chapter 2: ELEMENTS OF CELL ARCHITECTURE AND ORGANIZATION</p> <p>2.1. The cell of <i>Escherichia coli</i> – an organizational model of the prokaryotic cells</p> <p>2.2. The eukaryotic cell – dimensions, organization, components of the genetic apparatus</p> <p>2.2.1. General organization of the cell membrane</p> <p>2.2.2. The cytoskeleton</p> <p>2.2.3. The cytoplasm</p> <p>2.2.4. Cytoplasmic organelles with a role in the genetic apparatus of the cell: ribosomes, endoplasmic reticulum, Golgi apparatus, mitochondria, centrosome, lysosomes, peroxisomes</p>		2
3.	<p>Chapter 2: ELEMENTS OF CELL ARCHITECTURE AND ORGANIZATION</p> <p>2.2.5. Nucleus: nuclear envelope; nucleoplasm; nucleoplasmic nuclear bodies: nucleolus, Cajal body, histone locus body, promyelocytic leukemia oncoprotein body, nuclear spots; chromatin</p> <p>2.2.6. Metaphase chromosome: stages of its formation, morphology, morphological types, composition of karyotype, karyogram and idiogram</p> <p>2.2.7. Special types of chromosomes: polytene chromosomes and lampbrush chromosomes</p>		2

4.	<p>Chapter 3: ELEMENTS OF FUNDAMENTAL MOLECULAR GENETICS</p> <p>3.1. Chemical structure of nucleic acids</p> <p>3.2. Deoxyribonucleic acid (DNA): primary structure; secondary structure; tertiary structure; triplex DNA; quadruplex DNA; DNA methylation</p> <p>3.3. Ribonucleic acid (RNA): primary structure; secondary structure; tertiary structure; types of RNA, their synthesis and functions</p> <p>3.4. Physico-chemical properties of nucleic acids: DNA denaturation and the hyperchrome effect; DNA renaturation and the hypochromic effect; determining the concentration and purity of nucleic acid samples based on their property to absorb ultraviolet light; hydrolysis of nucleic acids</p>		2
5.	<p>Chapter 4: EVENTS OF CYTOGENETIC INTEREST DURING CELL DIVISIONS</p> <p>4.1. Interphase preparation within the cell cycle</p> <p>4.2. The possibilities of cell division</p> <p>4.3. Equal distribution of chromosome number during mitotic division</p>		2
6.	<p>Chapter 4: EVENTS OF CYTOGENETIC INTEREST DURING CELL DIVISIONS</p> <p>4.4. Reduction of diploid chromosomal number to haploid during gamete formation</p> <p>4.5. Segregation of mitotic chromosomes vs. segregation of meiotic chromosomes</p> <p>4.6. Gametogenesis through oogenesis and spermatogenesis</p>		2
7.	<p>Chapter 5: NUCLEAR INHERITANCE</p> <p>5.1. Mechanisms of autosomal inheritance of characters and traits</p> <p>5.1.1. Monogenic autosomal transmission</p> <p>5.1.1.1. Complete dominance and recessiveness: hybridological analysis and experiments with pea plants; the experiments of Gregor Mendel – between the controversies of the times and the path of the first Laws of Heredity; backcrossing and identifying the genetic structure of individuals with a dominant phenotype; the genomic era and knowledge of the characters investigated by Gregor Mendel in pea plants; complete dominance in human and animal pathology: oculocutaneous albinism, leukism, segregation of the Rh factor in humans and maternal-fetal incompatibility, bradydactyly and polydactyly, keratosis palmaris et plantaris, phenylketonuria, Huntington's syndrome, Tay-Sachs syndrome or GM2 gangliosidosis.</p>		2
8.	<p>Chapter 5: NUCLEAR INHERITANCE</p> <p>5.1. Mechanisms of autosomal inheritance of characters and traits</p> <p>5.1.1.2. Incomplete dominance</p> <p>5.1.1.3. Codominance</p> <p>5.1.1.4. Superdominance</p> <p>5.1.1.5. Lethality</p> <p>5.1.1.6. Pleiotropy</p> <p>5.1.1.7. Multiple alleles</p>	Power Point presentation, oral presentation	2
9.	<p>Chapter 5: NUCLEAR INHERITANCE</p> <p>5.1.2. Interactions between non-allelic genes to encode a character</p> <p>5.1.2.1. Complementarity</p> <p>5.1.2.2. Epistasis</p> <p>5.1.2.3. Polymeric and polygenic interactions: polymeric interaction in the coding of qualitative characters; polygenic inheritance of quantitative characters</p> <p>5.1.2.4. The modifying interaction of genes.</p>		2
10.	<p>Chapter 5: NUCLEAR INHERITANCE</p> <p>5.2. Mechanisms of heterosomal inheritance of characters and traits</p> <p>5.2.1. X-linked inheritance</p> <p>5.2.2. Z-linked inheritance</p> <p>5.2.3. Y- and W-linked inheritance</p> <p>5.3. Sex-linked characters vs. sex controlled characters</p> <p>5.4. The mechanism of linked inheritance of characters</p> <p>5.5. Penetrance and expressivity in nuclear inheritance</p>		2

11.	Chapter 5: NUCLEAR INHERITANCE 5.6. Elements of fundamental eredopathology in the mechanism of nuclear inheritance 5.6.1. Definition and classification of genetic diseases 5.6.2. Peculiarities of monogenic diseases inheritance 5.6.2.1. The inheritance of autosomal monogenic diseases 5.6.2.2. The inheritance of monogenic X-linked diseases	Power Point presentation, oral presentation	2
12.	Chapter 5: NUCLEAR INHERITANCE 5.6.3. Chromosomal disorders 5.6.3.1. Numerical chromosomal mutations 5.6.3.2. Chromosomal structural mutations, chromosomal deletion; chromosomal duplication; chromosomal inversion; other chromosomal structural mutations: transposition, translocation 5.6.3.3. Gene mutations 5.6.4. Chromosomes in oncogenesis		2
13.	Chapter 5: NUCLEAR INHERITANCE 5.6.5. Elements of genetic prophylaxis 5.6.5.1. Cytogenetic analysis 5.6.5.2. Pedigree analysis 5.6.5.3. Evaluation of heterozygosity status by test-cross		2
14.	Chapter 6: MITOCHONDRIAL INHERITANCE 6.1. The mitochondrial genome 6.2. Mitochondrial genome segregation 6.3. Justification of uniparental inheritance and the maternal effect in the expression of characters 6.4. Mitochondria and mitochondrial genetic diseases 6.5. Peculiarities in mitochondrial DNA decoding 6.6. The peculiarities of mitochondrial DNA in plants. Chloroplasts and chloroplastic DNA		2
TOTAL HOURS - Lectures			28

Bibliography

1. Grădinaru A.C., 2023 – *Medical Genetics and Eredopathology*, Lectures (*Writing in progress*).
2. Gupta P.K., 2007 – *Cytogenetics*, Rajsons Printers, New Delhi, India.
3. Hassan H., 2005 – *Mendel and the laws of Genetics*, The Rosen Publishing Group, New York.
4. Khanna P., 2009 – *Essentials of Genetics*, I.K. International Publishing House, New Delhi, India.
5. Russel P.J., 2006 – *Genetics: A Mendelian approach*, Pearson/Benjamin Cummings, San Francisco.

Crt. no.	8.2 Seminar / Laboratory/Practical work	Teaching methods	No. of hours
1.	Study methods used in Genetics	Power Point presentation	2
2.	Cellular elements with genetic role. Cytogenetic smear stages		2
3.	Metaphase chromosome morphology. Chromosome bands		2
4.	Segregation of chromosomes during cell divisions	Power Point presentation, activities of microscopy	2
5.	Mendelian genetics. Probability theory in Mendelian genetics. Simple and compound event probabilities in veterinary medical genetics. Backcross or backcross. <i>Case studies</i>	Case studies, analysis of segregation schemes, probabilities	2
6.	Mendelian genetics. The action and effect of allele genes. Types of interactions. Verification of Mendelian segregations by the Chi-square test. <i>Case studies</i>		2
7.	Mendelian genetics. The action and effect of non-alleles. Types of interactions. <i>Case studies</i>		2
8.	Morganism. <i>Drosophila melanogaster</i> – biological material in genetic studies. The phenomenon of gene linkage and crossing-over. Using crossing-over frequency to construct chromosomal genetic maps. <i>Drosophila</i> and <i>Abraxas</i> types of sexual determinism. Sex-linked Characters in <i>Drosophila</i> and <i>Abraxas</i> . Sex-controlled characters (limited or influenced by sex). <i>Case studies</i>		2

9.	Population genetics. Gene and genotype frequency in incomplete dominance. Genetic Hardy-Weinberg equilibrium of populations. <i>Case studies</i>	Case studies, analysis of segregation schemes, statistical calculation	2
10.	Population genetics. Gene and genotype frequency in complete dominance, multiple allelism and sex-linked genes. <i>Case studies</i>		2
11.	Evaluation of polygenic characters. Indicators of variability. Student's t test for analysis of variance. <i>Case studies</i>	Statistical calculation, genetic interpretation	2
12.	Evaluation of polygenic characters. Heritability and repeatability of characters		2
13.	Evaluation of polygenic characters. Standard Pearson correlation and phenotypic correlation		2
14.	Semester evaluation (40% of the final exam grade). Recovery of absent practical works		2
TOTAL HOURS – Practical works			28

Bibliography

1. Grădinaru A.C., 2023 – *Medical Genetics and Eredopathology, Practical work notes (Writing in progress)*.
2. Grădinaru A.C., 2022 – *Medical Genetics and Eredopathology, Lecture notes, (Writing in progress)*.
3. Gupta P.K., 2007 – *Cytogenetics*, Rajsons Printers, New Delhi, India;
4. Hassan H., 2005 – *Mendel and the laws of Genetics*, The Rosen Publishing Group, New York;
5. Khanna P., 2009 – *Essentials of Genetics*, I.K. International Publishing House, New Delhi, India;
6. Russel P.J., 2006 – *Genetics: A Mendelian approach*, Pearson/Benjamin Cummings, San Francisco.

9. Corroborating the contents of the subject with the expectations of community representatives, professional associations and representative employers in the field of the program

The content of the subject provides skills in cytogenetic and molecular diagnosis, autosomal and heterosomal segregation and monogenic inheritance, as well as polygenic inheritance of characters.

10. Evaluation

Activity type	Evaluation criteria	Methods of evaluation	Share from the final grade (%)
10.1 Lecture	Knowing the syllabus content for lectures	Written exam	60
10.2 Seminar /laboratory activities	Knowing the syllabus content for practical activities	Written evaluation	40

11. Standards of performance

Minimum requirements (for grade 5): Knowing basic mechanisms of Cytogenetics and Molecular Genetics involved in characters and traits encoding and expressing processes. Interpreting the results of segregation schemes for various characters. Knowing the genetic substrate of various congenital diseases.	Maximum requirements (for grade 10): Proper knowledge of Genetics including discussions about various traits, genetic diseases, their mechanisms of inheritance, genetic diagnosis in Veterinary Medicine, ways of Genetic population structure establishing.
--	---

Signature of *Lectures* coordinator
Assoc. Prof. Andrei C. GRĂDINARU

Signature of *Practical Applications* coordinator
Assoc. Prof. Andrei C. GRĂDINARU

Signature of the *Head of the Department*
Asoc. Prof. Geta PAVEL

11.09.2023

Approved in *Preclinics Department*:
15.09.2023