UNICAMILLUS

Degree in Medicine and Surgery
Integrated Course of Biology and Genetics
CFU:10
Professor responsible: **Laura Pacini**

Discipline: **Applied Biology**
SSD: **BIO/13**
CFU: **9**

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Discipline: **Medical Genetics**
SSD: **MED/03**
CFU: **1**

Professor: **Cinzia Ciccacci** mail: **cinzia.ciccacci@unicamillus.org**

PREREQUISITES

Not applicable for Biology. For Medical Genetics it would be desirable that the student already knows basic biology elements, such as the gene structure, DNA replication, concepts of mitosis and meiosis.

LEARNING OUTCOMES

The integrated course of Biology and Genetics aims to provide students with the functional logic of living systems, with particular attention to the properties and functions of the cell as a basic unit of life. The student will learn the unitary mechanisms that regulate the processes and activities of the cell and the interactions between cells; the principles that govern the biological units diversity, in relation to their structural and functional characteristics, to the modalities of gene expression both within the different districts of a single individual (differentiation), and longitudinally, during evolution.
The fundamental principles of molecular biology and genetics will also be addressed; particular emphasis will be given to aspects useful to students in medicine, such as the cellular and molecular bases of diseases and the effects of drugs on cell structure and function. The Medical Genetics part will provide students the knowledge on the main notions on the inheritance of monogenic, chromosomal and multifactorial diseases. At the end of the course, the student will be able to distinguish the main classes of genetic diseases and to recognize the modes of transmission of hereditary diseases.

Knowledge and understanding: Knowledge of the fundamentals of molecular and cellular biology: the student will acquire the general and unitary principles governing living organisms' functions and behavior, including the mechanisms that operate in the transmission of hereditary traits.

At the end of this course the student will be able to:

- know the principles of classification of living organisms.
- Describe the main characteristics and differences between prokaryotic and eukaryotic cells.
- Know the main cellular compartments and their function.
- Know the general principles of cellular metabolism.
- Know the molecular basis of transmission of hereditary traits.
- Know the molecular basis of gene expression.
- Know the differences between mitosis and meiosis.
- Know the basic mechanisms of cell differentiation.
- Recognize and describe the dysregulation consequences of the main processes operating in the cell and the molecular bases of the diseases.
- Know the correct genetic terminology.
- Know the main inheritance models of monogenic, chromosomal and multifactorial diseases.
- Know the main biological mechanisms that cause hereditary diseases.
- Reconstruct family pedigrees and calculate disease recurrence.
- Comprehend and use the main kinds of genetic tests properly.

Applying knowledge and understanding: The general objective of the integrated biology and genetics course is to learn the experimental method and its applications to the study of fundamental biological phenomena. Therefore, the course aims to arouse the ability to perform precise and documented observations and make a correct critical analysis to derive verifiable generalizations. At the end of the course, the student will be able to apply the experimental method to the study of biological processes. Moreover, the student will acquire the tools to understand and explain the molecular and cellular mechanisms that are the basis of diseases. He will also be able to analyse family pedigrees and clinical and molecular genetic data useful for genetic counselling
and to calculate disease recurrence risk.

Communication skills: The student will be able to adequately describe a biological phenomenon by demonstrating that he/she has learned an appropriate scientific language for correct and rigorous communication. He/she will be able to describe the main models of inheritance and the recurrence risk the using a correct genetic terminology.

Making judgements: At the end of the course, the student will have analyzed and learned the exemplary biology experiments and will be able to develop autonomously the logical procedures and strategies that allow to apply the experimental method, analyze and correctly interpret experimental data.
Furthermore, he will have acquired the ability to synthesize and correlate the various topics and to critically use genetic tests for the molecular diagnosis of monogenic and chromosomal diseases or for the evaluation of genetic susceptibility to complex diseases.

Learning skills: The student will have acquired skills and learning methods suitable for the deepening and improvement of their skills in biology. At the end of the course, the student will have developed the ability to deepen the topics through the consultation of the scientific literature.

SYLLABUS

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BIOLOGY

Characteristics of living organisms. Cell theory, the cell as a structural and functional unit of life.

Chemistry of life: Macromolecules: carbohydrates, lipids, proteins and nucleic acids (structure, shape and function).

Prokaryotic and eukaryotic cells: classification and main structural differences. Organization of the eukaryotic cell. Cellular organelles (structure and function). Notes on viruses as endocellular parasites.

Plasma membrane. The fluid-mosaic model of the cell plasma membrane. Main functions of membrane proteins and their topological organization in the lipid bilayer. Modes of ions and small molecules transport across the plasma membrane (simple diffusion, facilitated diffusion, active transport). The basis of membrane excitability.

Compartmentalization in the eukaryotic cell. The cytoplasm and the system of endocellular membranes: structure and function of endoplasmic reticulum, Golgi apparatus and lysosomes. Notes on peroxisomes.

Mitochondria and chloroplasts. Structure and function of mitochondria and chloroplasts as generators of energy. Notes on glycolysis, fermentation and cellular respiration. The endosymbiotic theory of the origin of mitochondria and chloroplasts.

Cytoskeleton, adhesion and cell motility. The cytoskeleton. Structure and function of intermediate filaments, microtubules, and actin filaments. Molecular motors. Cellular structures that determine the shape, polarity, and motility of the cell. Interactions between cells and their environment. The molecules adhesion molecules and the extracellular matrix.

Nuclear compartment. Nuclear envelope, nucleolus, organization and different levels of chromatin condensation, chromosomes.

Molecular bases of hereditary information. DNA structure and function. Identification of DNA as genetic material. Molecular mechanism of DNA duplication. Telomeres and Telomerase. Mechanisms of DNA repair, correlations with human pathologies.

RNA structure and function. Main types of RNA present in the prokaryotic and eukaryotic cell.
RNA Transcription and RNA processing in eukaryotic cells, with particular attention to the maturation of the messenger RNAs. Role of non-coding RNAs.

Protein synthesis. Ribosomes: structure and biological role, differences between prokaryotic and eukaryotic ribosomes. Properties of the genetic code, general features of translation and biological implications.

Post-synthetic fate of proteins. Post-translational modifications and fate of proteins after synthesis. Signals and mechanisms of protein sorting to organelles and secretory pathway.
Functions of the endoplasmic reticulum in the protein sorting (signal and stop sequences). Endoplasmic reticulum and Golgi apparatus role in protein glycosylation.

Vesicle trafficking. Modes of protein transport between different cellular compartments. Endocytosis (pinocytosis, phagocytosis, receptor-mediated endocytosis); autophagy; Constitutive and regulated exocytosis.

Control of gene expression. Molecular mechanisms that create specialized cell types. Functional organization of the eukaryotic genome. Histone code.

Control at the transcriptional level in prokaryotic and eukaryotic cells. Role of chromatin condensation state and degree of DNA methylation (epigenetic modifications). Main strategies of post-transcriptional and post-translational control.
Mitosis and Meiosis. Principles of chromosome dynamics during mitosis and meiosis, differences between the two processes. Genetic consequences of meiosis, the importance of meiosis as a source of genetic variability. Molecular mechanisms of genetic recombination. Concept of haploidy and diploidy. Homologous chromosomes. Characteristics of sexual and asexual reproduction.
Cellular communication and signal transduction. Communication between cells in multicellular organisms. General principles of cell signaling, chemical signals and receptor proteins.
Mechanisms of signal transduction and main signaling pathways. Nuclear receptors, G-protein coupled receptors, Enzyme-coupled receptors. Second messengers. Protein kinases and molecular switches.
Cell Cycle, mechanisms of cell death. Cell cycle, phases of the cycle and control of progression along the cell cycle as a result of the interaction between intracellular mechanisms and extracellular signals. The role of cyclin-dependent kinases.
Basic knowledge of the processes of apoptosis, necrosis and autophagy.
Molecular basis of cancer. Molecular mechanisms of tumor transformation. Characteristics of the neoplastic cell. Tumor suppressors and proto-oncogenes. Genetic and epigenetic alterations underlying tumors.

MEDICAL GENETICS
Basic Genetics: Definitions of Key Terms: gene, locus, allele, genotype, phenotype, haplotype, homozygous, heterozygous, haploid, diploid, dominance, recessivity, codominance, mutation, polymorphism.
Principles of Genetic Transmission: Mendel's Genetic Hypothesis, The Monohybrid and Dihybrid Crosses, Segregation in Human Pedigrees, Blood groups Genetics
Monogenic Inheritance Models: Autosomal inheritance, Autosomal recessive inheritance, X-linked inheritance
Genetic Risk calculation and pedigrees analyses.
Population Genetics: Hardy-Weinberg equilibrium and theoretical implications for understanding the mechanisms of biological evolution.
Chromosomes: Structure and Analysis, Chromosomes Pathologies. Genomic Imprinting.
X-chromosome inactivation
Mitochondrial inheritance: mitochondrial DNA, pattern of inheritance
Multifactorial Inheritance and Common Diseases: polimorphisms, susceptibility genes, gene environment interaction, association studies.
Pharmacogenomics and Personalised Medicine
Genetic tests and Counselling. Outlines

COURSE STRUCTURE

The course is divided in lectures, 90 hours Biology and 10 hours Medical Genetics. The professor uses educational tools such as presentations organized in powerpoint files with explanatory diagrams, illustrations and images which are taken from micrographies to describe the various cellular structures, and movies and animations to complement the processes described in class.
The possibility of ongoing tests is provided for. Lectures on medical genetics will include theoretical lessons with power-point presentations and exercises (both in groups and alone).
The attendance at lectures is mandatory.

COURSE GRADE DETERMINATION

Student learning will be assessed through a written tests in biology and genetics, and an oral exam on the biology module, held at the end of the course.
The written test is mandatory to be admitted to the oral exam.
The written test is organised into 30 questions for Biology and 30 questions for Medical Genetics with multiple-choice answers in which only one is correct. Each correct answer is given a score of 1 for Biology and 1 for Medical Genetics. There is no penalty for wrong or not given answers. To access the oral test, the student must have obtained a grade $\geq$ 18/30 in both the Biology and Medical Genetics modules. Notably, the written test is a selection test; in the oral exam the student is given the opportunity to demonstrate his preparation by discussing the topics of the course, to reason on issues related to biology by showing that he/she has acquired the ability to express himself in a suitable scientific language.
The final grade will result from a weighted average between the grade on the biology exam and the grade on the written genetics test.

READING MATERIALS

"Molecular Biology of the Cell", Bruce Alberts et al., VII ed., WW Norton & Co.
"Essential Cell Biology", Bruce Alberts et al. V ed., WW Norton & Co.
"Medical Genetics" by Lynn Jorde, John Carey, Michael Bamshad. Edited by Elsevier