UNICAMILLUS

Degree in Medicine and Surgery

Integrated Teaching of Biology and Genetics
SSD: BIOS-10/A, MEDS-01/A
Coordinator: **Roberta Nardacci** mail: **roberta.nardacci@unicamillus.org**
Total CFU:**9**

Module: **Applied Biology**
SSD: **BIOS-10/A**
Professor: **Roberta Nardacci** **(1 CFU)** mail: **roberta.nardacci@unicamillus.org**
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CFU: **8**

Module: **Medical Genetics**
SSD: **MEDS-01/A**
Professor: **Andrea Latini** **(1 CFU)** mail: **andrea.latini@unicamillus.org**
CFU: **1**

PREREQUISITES

Not applicable for the Biology module. For the Medical genetics module it is recommended that students have a solid background in biology, particularly in understanding gene structure, the DNA replication process and the concepts of mitosis and meiosis.

LEARNING OUTCOMES

The integrated teaching 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 cell processes and activities as well as interactions between cells; the principles governing the diversity of biological units, in relation to their structural and functional characteristics as well as modes of gene expression. This analysis will occur in both the context of individual differentiation and within the framework of evolution.

Fundamental principles of molecular biology and genetics will also be addressed; special emphasis will be placed on aspects relevant to medical students, such as the cellular and molecular bases of diseases and the effects of drugs on cell structure and function. The Medical Genetics module will provide key knowledge about the inheritance of monogenic, chromosomal and multifactorial diseases. By the end, the student will have acquired the main analysis method for diagnosing genetic diseases, will be able to distinguish the major classes of genetic diseases and recognize their modes of transmission.

Knowledge and understanding: Understanding of the fundamentals of molecular and cellular biology. The student will acquire general and unitary principles governing the functioning and behavior of living organisms including the mechanisms involved in the transmission of hereditary traits.

At the end of this integrated teaching the student will be able to:
- Define a living organism.
- Recognize the common characteristics of all living organisms.
- Describe the 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.
- Understand the molecular basis of gene expression and its regulation.
- 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.

Applying knowledge and understanding: The overall goal of the integrated biology and genetics teaching is to learn the scientific method and its applications in the study of fundamental biological phenomena. Therefore, the main objective will be to stimulate the ability to conduct accurate and well documented observations as well as to develop critical and analytical skills in order to make verifiable generalizations. By the end of the course, students should be able to apply the scientific method to the study of biological processes and understand and explain the molecular and cellular mechanisms underlying diseases. Additionally, students will acquire the ability to analyze family pedigrees and clinical and molecular genetic data useful for genetic counseling. Finally, they will be able to calculate recurrence risk of genetic diseases.

Communication skills: The student will be able to properly describe a biological phenomenon by demonstrating that they have learned an appropriate scientific language for correct and rigorous communication. They will be able to describe the main patterns of inheritance and the recurrence risk using correct genetic terminology.

Making judgments: At the end of the course, the student will have analyzed and learned exemplary biology experiments, and will be able to autonomously develop the logical procedures and strategies required for the application of the experimental method as well as for the analysis and the correct interpretation of the experimental data.

Furthermore, he/she will acquire the necessary skills required for the synthesis and correlation of different topics and will develop the ability to recognize the main inheritance models and will be able to attribute them to monogenic, chromosomal or multifactorial diseases.

Learning skills: The student will acquire skills and learning methods suitable for the deepening and the advancement of their knowledge in the field of Biology and Medical Genetics, especially through consultation of scientific literature.

SYLLABUS

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 intracellular 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).
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. Overview on DNA repair.
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.
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.
Compartmentalization in the eukaryotic cell. The cytoplasm and the system of intracellular membranes: structure and function of endoplasmic reticulum, Golgi apparatus and lysosomes. Notes on peroxisomes.
Post-synthetic fate of proteins. Post-translational modifications and fate of proteins after synthesis.

Cell trafficking and protein localization. Signals and mechanisms of protein sorting to organelles and secretory pathways.

Functions of the endoplasmic reticulum in the protein sorting (signal and stop sequences). Endoplasmic reticulum and Golgi apparatus role in protein glycosylation.

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.

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

Mitosis and Meiosis. 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.

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**.**

Mechanisms of cell death. Basic knowledge of the processes of apoptosis, necrosis and autophagy.

Vesicle trafficking. Endocytosis (pinocytosis, phagocytosis, receptor-mediated endocytosis); autophagy; Constitutive and regulated exocytosis.

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. The basis of membrane excitability: Neurons and action potential.

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, 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. **X-chromosome inactivation**

Mitochondrial inheritance

Multifactorial Inheritance and Common Diseases: polimorphisms, susceptibility genes, gene environment interaction, association studies.

COURSE STRUCTURE

The course is divided into classroom lectures, of which 80 hours dedicated to Biology and 10 hours dedicated to Medical Genetics. The instructors use various educational tools including organized PowerPoint presentations with explanatory diagrams, illustrations and images which are taken from micrographies to describe the various cellular structures. Videos and animations will be employed to complement the topics discussed in class. Interactive lessons with in-class exercises (both individual and group-based) are planned for the Genetic module. Intermediate tests may be scheduled for both the modules.

The attendance at lectures is mandatory with a minimum requirement of at least 67% attendance of the total hours scheduled for all integrated course lectures.

COURSE GRADE DETERMINATION

Student learning will be assessed through a written test and an oral exam held at the end of the course. The written test is mandatory to be admitted to the oral exam.

The written test is organized into 60 questions with multiple-choice answers in which only one is correct. Each correct answer is given a score of 0,5. 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 the written test. Notably, 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.

Hence, the whole examination will be evaluated as it follows:

- **Insufficient**: severe poor knowledge of the subject, very limited skill in the analysis of specific items.
- **18-20**: knowledge of the subjects of sufficient quality characterized by frequent imperfections. Analysis and reasoning skills of sufficient quality.
- **21-23**: standard knowledge of the specific subject; analysis and reasoning skill of acceptable quality.
- **24-26**: good knowledge of the subjects and good analysis and reasoning skills; arguments are expressed in a rigorous way.
- **27-29**: very good knowledge of the specific scientific subjects, valid analysis and reasoning skills, significant skill in making judgements.
- **30-30L**: outstanding knowledge of the specific knowledge of the scientific tasks. Exceptional analysis, reasoning and making judgments skills.

READING MATERIALS

The recommended textbooks should be considered as reference teaching material. However students may choose the text(s) they prefer or deem most suitable for their learning needs. Additional material will be provided by the lecturer.

- “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”, Lynn Jorde, John Carey, Michael Bamshad. Edited by Elsevier.