DEGREE IN MEDICINE AND SURGERY

Integrated Course of Biology and Genetics

Discipline : **Applied Biology**

SSD : **BIO/13**

CFU : **9**

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Professor: **Emiliano Maiani (1cfu)** e-mail : **emiliano.maiani@unicamillus.org**

Discipline : **Medical Genetics**

SSD : **MED/03**

CFU : **1**

Professor: **Cinzia Ciccacci** e-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 meiosis and mitosis.

LEARNING OUTCOMES

The integrated course in 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 diversity of biological units, in relation to their structural and functional characteristics, to the modes 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 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 the functions and behavior of living organisms 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 beings.
- 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 consequences of a dysregulation of the main processes that operate in the cell
- 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 properly the major kinds of genetic tests.

Applying knowledge and understanding: The general objective of the integrated course in biology and genetics 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 to make a correct critical analysis in order 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 and will have acquired 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 the purposes of correct and rigorous communication. He 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 experiments of biology 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

Biology

Characteristic of living cells: Cellular theory. Classification principles of living organisms.

Cell Chemistry: Macromolecules: structure, shape and function.

Prokaryotic and eukaryotic cell models: classification and major structural differences, Organelles (structure and function). Eukaryotic nuclear compartment, structure and functions.

Plasma membrane: properties and functions, modes of transport of small molecules across the plasma membrane (simple diffusion, facilitated diffusion, active transport).

Internal organization of the cell: intracellular compartments and protein sorting, intracellular membrane traffic, organelles. The cytoskeleton and cell motility. Nuclear compartment, structure and functions. The different levels of chromatin condensation.

Energy conversion: Glycolysis and fermentation (outline). Mitochondria and Chloroplasts, structure and function.

Molecular basis of hereditary information: DNA replication. DNA repair and its correlation with human diseases.

RNA, structure and function: Main types of cellular RNAs and differences with respect to DNA in terms of molecular size, stability and biological functions.Transcription and RNA maturation.

Genetic Code and its properties. Protein synthesis: How cells read the genome. Main post-translational modifications of the polypeptide chains.

Post-synthetic fate of proteins, endomembranes and membrane traffic: Endocytosis, pinocytosis, phagocytosis and receptor-mediated endocytosis (LDL).

Control of gene expression: Molecular mechanisms that create specialized cell types. Functional organization of the eukaryotic genome. Histone code. Transcriptional control, role of chromatin condensation and of the degree of DNA methylation.

Cell Cycle: The cell cycle control system. Apoptosis and Cancer, Tumor suppressors and proto-oncogenes.

Cell Signaling: Exchange of chemical signals through receptor proteins. The key role played by protein kinases in this process.

The interactions between cells and their environment: Adhesion molecules and extracellular matrix.

Mitosis and Meiosis: principles of chromosome dynamics during mitosis and meiosis; molecular mechanisms of genetic recombination.

Gene mutations: mutations by substitution, insertion or deletion of nucleotides. Spontaneous and induced mutations. Chemical and physical mutagens. DNA repair systems for single or double stranded DNA damage.

Mobile genetic elements and evolution of the genomes

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 frontal lectures, 90 hours (80 Hours Biology and 10 hours Medical Genetics), the teacher uses educational tools such as presentations organized in powerpoint files with explanatory diagrams, illustrations and images taken from micrographies to describe the various cellular structures and movies and animations to complement the processes described in class. The attendance at lectures is mandatory.

The possibility of ongoing tests is provided for.

Lectures of medical genetics will include theoretical lessons with power-point presentations and exercises (both in groups and alone).

COURSE GRADE DETERMINATION

Student learning will be assessed through a written and oral exam held at the end of the course.

The written test is mandatory to be admitted to the oral exam, but it does not contribute to the final score. The written test is organised into 30 questions for Biology and 15 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 2 for Medical genetics. There is no penalty for wrong or not given answers.

To access the oral test, the student must have obtained at least 18/30 in both the Biology and Medical Genetics modules. Notably, the written test is a selection test; in the oral test the student is given the opportunity to demonstrate his preparation by discussing the topics of the course, to reason on issues related to biology and genetics by demonstrating that he has acquired the ability to express himself in a suitable scientific language. The final evaluation will be based mainly on the outcome of the oral test.

READING MATERIALS

Molecular Biology of the Cell", VI edition, Bruce Alberts et al., Garland Science, 2014.

"Medical Genetics" by Lynn Jorde, John Carey, Michael Bamshad. Edited by Elsevier