

Degree Course of Physiotherapy

Integrated Teaching: BIOLOGY, BIOCHEMISTRY AND GENETICS

NUMERO DI CFU:6

SSD: BIO/10, BIO/13, MED/03

Coordinator: CINZIA CICCACCI

e-mail: cinzia.ciccacci@unicamillus.org

MODULE: BIOCHEMISTRY

CFU: 2

SSD: BIO/10

Professor: PROF. GIAMPIERO MEI

e-mail: giampiero.mei@unicamillus.org

MODULE: MEDICAL GENETICS

CFU: 2

SSD: MED/03

Professor: PROF.SSA CINZIA CICCACCI

e-mail: cinzia.ciccacci@unicamillus.org

MODULE: APPLIED BIOLOGY

CFU: 2

SSD: BIO/13

Professor: PROF. Roberta Nardacci

e-mail: roberta.nardacci@unicamillus.org

PPREREQUISITES

BIOCHEMISTRY

Knowledge is required of some general notions of chemistry (atomic / molecular structure, valence, pH, molar concentration).

MEDICAL GENETICS

There are no prerequisites, but it should be better if student already knows basic biology elements, such as the gene structure, DNA replication, concepts of meiosis and mitosis.

LEARNING OBJECTIVES

BIOCHEMISTRY

At the end of the course the student must be able to identify the states of the matter and the significance of the chemical solutions and concentrations; it must also be able to know the organic and inorganic composition of the human organism, the general characters and the biological role of the main elements of organic chemistry.

MEDICAL GENETIC

The course aims to provide the student with the main notions on the inheritance of monogenic, chromosomal and multifactorial diseases. The student must acquire knowledge of the main methods of analysis useful for the diagnosis of these pathologies. It will also have to demonstrate the ability to analyse genealogical trees and clinical and genetic-molecular data for genetic counselling purposes

APPLIED BIOLOGY

The Biology module aims at providing the students with the morphological and functional organization of prokaryotic and eukaryotic cells, focusing on both the descriptive aspects and the basic notions of biochemistry and cellular physiology required to understand the functions of the cell as a basic unit of living organisms.

The objective of the course is the learning of the constructive logic of the biological structures at the different levels of organization of living matter, the principles that govern the functioning of the different biological systems, the learning of the experimental method and its applications to the study of biological phenomena.

Students will learn the unitary mechanisms that regulate cellular activities, gene expression and genetic material transmission.

LEARNING OUTCOMES

BIOCHEMISTRY

Knowledge and understanding

The students involved are identified in the average between the lessons and the exercises. What are the structures of the most important biological macromolecules. We will also have to demonstrate, through the final test, that we know how to investigate the role of these molecules in the context of the main metabolic processes that take place in the cell.

Applying knowledge and understanding

Students will be repeatedly tested through open questions on quantitative (numerical) and qualitative biochemistry problems (for example inherent diseases related to dysfunctions / deficiencies of which the molecular origin is known) on the topics covered in class, in order to evaluate them constantly study skills.

Making judgements

During the lessons, students will also be asked questions whose answers require, starting from the knowledge acquired, a reasoning of logic (type cause-effect). In this way students will be induced to think autonomously, each evaluating their own deductive skills in the subsequent collegial discussion of the answers given.

MEDICAL GENETIC

Knowledge and understanding

At the end of the course, the student must have acquired:

- The knowledge of correct genetic terminology
- The knowledge of the main inheritance models of monogenic, chromosomal and multifactorial diseases
- The knowledge of the main biological mechanisms that cause hereditary diseases
- The understanding of how to reconstruct family pedigrees and to calculate disease recurrence
- The understanding of the major kinds of genetic testing and their proper use.
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Applying knowledge and understanding

- The capacity to analyse family pedigrees and clinical and molecular genetic data useful for genetic counselling;
- The ability to calculate disease recurrence risk.

Communication skills

- The use of correct genetic terminology;
- The ability to describe the main models of inheritance and the recurrence risk.

Making judgements

The ability to synthesize and correlate the various topics. A critical ability on the use of genetic tests for the molecular diagnosis of monogenic and chromosomal diseases or for the evaluation of genetic susceptibility to complex diseases.

APPLIED BIOLOGY

Knowledge and understanding: At the end of the course the student will learn :

- The classification principles of living organisms.
- The major structural differences between prokaryotic and eukaryotic cells.
- Describe the cellular morphology.
- Describe the cellular compartments and their function.
- Know the general principles of cellular metabolism.
- Know the molecular basis of hereditary information.
- Know the differences between mitosis and meiosis.

Applying knowledge and understanding: The course aims to provide students with the knowledge and methodological tools for the study of the biological bases of life, the knowledge of the experimental method and its applications to the study of fundamental biological phenomena.

Making judgements: Students will be able to develop autonomously the logical processes and strategies that allow to apply the experimental method, formulate a scientific hypothesis, analyze and correctly interpret experimental data.

Communication skills: Students will be able to describe a biological phenomenon and demonstrate that they have learned an appropriate scientific language for the purposes of correct and rigorous communication.

Learning skills: The student will acquire skills and methods of learning suitable for the deepening and updating of their proficiency in biology

COURSE SYLLABUS

BIOCHEMISTRY

A – FUNDAMENTALS OF CHEMISTRY: Atomic structure. Definition of oxides, acids, bases and salts. Examples. The valence. Covalent and hydrogen bonds. The properties of water. Osmosis. pH. Molar concentration of solutes. The fundamental chemical groups of organic molecules.

B – FUNDAMENTALS OF STRUCTURAL BIOCHEMISTRY: Proteins structure and function. Enzymes. Carbohydrates. Fatty acids. Vitamins and hormones. Nucleic acids.

C – FUNDAMENTALS OF METABOLIC PROCESSES: Reducing power: NADH, NADPH, FADH₂. Introduction to metabolism: Glycolysis. Krebs cycle. Fatty acid catabolism. The respiratory chain.

MEDICAL GENETICS

Basic concepts and terminology: gene, locus, allele, genotype, phenotype, haplotype, homozygous, Heterozygote, haploid, diploid, dominance, recessivity, codominance, mutation, polymorphism.

Laws of Mendel

Inheritance models of Mendelian (or monogenic) characters: autosomal recessive and dominant inheritance, inheritance linked to recessive and dominant sex.

The genetics of the main blood groups (AB, Rh). Fetal maternal incompatibility

Repeated triplet diseases or dynamic mutations

Risk calculations related to the above mentioned models and family tree analysis

Concepts of penetrance, expressivity, Epistase, anticipation, consanguinity, genetic heterogeneity

Chromosomes: structure and characteristics. Anomalies of number and structure of chromosomes. Chromosome study techniques

Genomic imprinting. Overview

X chromosome inactivation

Genetic markers and polymorphisms. Inter-individual genetic variability. Notes of Multifactorial Inheritance

Genetic Tests and their applications. Hints of genetic counseling.

Notes on pharmacogenetics and the concept of personalized medicine

Genetics of Neurological Diseases: some examples

APPLIED BIOLOGY

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

Prokaryotic and eukaryotic cell models: classification and major structural differences.

Cell Chemistry: Macromolecules: structure, shape and function

Plasma membrane: properties and functions.

Internal organization of the cell: Cellular compartments. Cytoplasm and cytoplasmic organelles, ribosomes, smooth and rough endoplasmic reticulum, Golgi apparatus, lysosomes, peroxisomes.

The cytoskeleton. Microtubules, intermediate filaments and microfilaments. Cilia and flagella.

Centrioles and centrosomes.

Energy conversion: Glycolysis, fermentation, cellular respiration, photosynthesis. (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 and exocytosis.

Cell Cycle, Mitosis and Meiosis

COURSE STRUCTURE

The teaching is structured in 2 CFU with 20 hours of frontal lessons. Lectures 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 exam held at the end of the course, in which the student answers to multiple choice questions on topics presented during lectures. The written test will consist of 30 questions with multiple choice answers, for each correct answer a 1 point will be assigned. The final score of the written test will be given by the sum of the partial scores assigned to correct answers. Oral exam is optional. To access the oral exam student must have obtained at least a minimum of 15 points at the written exam (15/30). The minimum score to pass the exam is 18/30.

READING MATERIALS

MEDICAL GENETICS

Lectures in pdf format will be provided to students.

Recommended books: "Medical Genetics" by Lynn Jorde John Carey Michael Bamshad. Edited by Elsevier

BIOCHEMISTRY

Lippincott Illustrated Reviews: Biochemistry (Lippincott Illustrated Reviews Series) Seventh, North American Edition, by Denise Ferrier

Lehninger Principles of Biochemistry, by D. L. Nelson and M. M. Cox

APPLIED BIOLOGY

- Sadava, Hillis, Heller, Hacker. Elementi di Biologia e Genetica Zanichelli editore, V ed.
- Raven, Johnson, Mason, Losos, Singer. Elementi di Biologia e Genetica Piccin editore II ed