Degree Course of Physioterapy

INTEGRATED COURSE: BIOCHEMISTRY BIOLOGY APPLIED GENETICS
CFU:6
SSD: BIO/10, BIO/13, MED/03
COORDINATOR: CINZIA CICCACCI MAIL cinzia.ciccacci@unicamillus.org

MODULE: BIOCHEMISTRY
CFU: 2
SSD: BIO/10
PROFESSOR: PROF. LUCA FEDERICI MAIL luca.federici@unicamillus.org
RECEIVING HOURS: by appointment

MODULE: MEDICAL GENETICS
CFU: 2
SSD: MED/03
PROFESSOR: PROF.SSA CINZIA CICCACCI MAIL **cinzia.ciccacci@unicamillus.org**
RECEIVING HOURS: by appointment

MODULE: APPLIED BIOLOGY
CFU: 2
SSD: BIO/13
PROFESSOR: PROF. Roberta Nardacci **MAIL:** roberta.nardacci@unicamillus.org
RECEIVING HOURS: by appointment

PREREQUISITES

BIOCHEMISTRY

Knowledge is required of some general notions of chemistry (atomic / molecular structure, valence, pH, molar concentration). The main basic notions required will in any case be recalled at the beginning of the course.

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

Knowledge of the main classes of organic compounds and biological macromolecules.
Knowledge of the functioning of respiratory proteins. Knowledge of enzymes functioning and kinetics.
Knowledge of the logic of energy metabolism in humans and the role played by the main classes of biomolecules.
General knowledge of the main metabolic pathways and, in more detail, of the glucose catabolism pathways.

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

Students will have to demonstrate a good knowledge of the structure and function of the most important biological macromolecules, describing also their role in the frame of the main metabolic processes that take place in the cell.

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.*

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

Recalls to the basic notions of general chemistry

The structure of the atom. Chemical bonds. The main properties of aqueous solutions: measurement of concentration, pH, acid-base properties. Thermodynamics and chemical equilibrium. The chemical kinetics. The redox reactions.

The chemistry of carbon

Hybridization of orbitals: sp, sp2, sp3. Aliphatic and aromatic hydrocarbons. Chirality. Alcohols, aldehydes, ketones and carboxylic acids. Amines, esters, anhydrides and amides. Redox reactions in organic chemistry.

Structure and function of biological molecules

Carbohydrates: general aspects and classification, monosaccharides, oligosaccharides, polysaccharides.

Lipids: general aspects and classification, fatty acids, acylglycerols, phosphoglycerides and sphingolipids, steroids. The biological membranes.
Amino acids and the structure of proteins: amino acids; peptide bond; primary structure; tertiary and quaternary secondary school.
Functions of proteins: Myoglobin and hemoglobin.
Enzymes: characteristics and functioning; mechanisms of enzymatic inhibition.
Carbohydrate metabolism: glycogen, glycolysis and gluconeogenesis. Hormonal regulation of blood sugar.
Lipid metabolism: fatty acids as main fuels of metabolism, beta oxidation, ketone bodies, synthesis of fatty acids.
Amino acid metabolism: digestion of proteins; transamination, deamination and urea production.
Bioenergetics: citric acid cycle, respiratory chain, electron transfer and synthesis of ATP through ATP synthase.

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. Hardy-Weinberg Equilibrium
Chromosomes: Structure and Analysis, Chromosomes Pathologies
Genomic Imprinting
X-chromosome inactivation
Mitochondrial inheritance: mitochondrial DNA, pattern of inheritance
Multifactorial inheritance: polimorphisms, susceptibility genes, gene-environment interaction, association studies
Pharmacogenomics and Personalised Medicine
Dynamic mutations and related disorders
Genetic tests and Counselling. Outlines

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 rought 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 6 CFU with 60 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.
The evaluation criteria adopted will be the following:
Unsuitable: Poor or lacking knowledge and understanding of the topics; limited capacity for analysis and synthesis, frequent generalizations of the required contents; inability to use technical language.
18-20: Just enough knowledge and understanding of topics, with obvious imperfections; just sufficient capacity for analysis, synthesis and independent judgement; poor ability to use technical language.
21-23: Sufficient knowledge and understanding of topics; sufficient capacity for analysis and synthesis with the ability to logically and coherently argue the required contents; sufficient ability to use technical language.
24-26: Fair knowledge and understanding of the topics; discrete capacity for analysis and synthesis with the ability to rigorously argue the required contents; Good ability to use technical language.
27-29: Good knowledge and understanding of required content; good capacity for analysis and synthesis with the ability to rigorously argue the required contents; good ability to use technical language.
30-30L : Excellent level of knowledge and understanding of the required contents with an excellent capacity for analysis and synthesis with the ability to argue the required contents in a rigorous, innovative and original way; Excellent ability to use technical language.

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

-"Biochemistry", D. R. Ferrier Wolters Kluwer;
- "Lehningher principles of biochemistry", D. L. Nelson, M.M. Cox (2017) W.H. Freeman & Co.

APPLIED BIOLOGY

- "Essential Cell Biology (Fifth edition)" by Bruce Alberts, Karen Hopkin, Alexander D Johnson, David Morgan, Martin Raff, Keith_Roberts, Peter Walter. Editor: W.W. NORTON

- "Biology" by Peter H. Raven, George Johnson, Kenneth A. Mason, Jonathan B. Losos, Tod Duncan. Editor: McGraw-Hill Education

The indicated textbook is just a reference. Students are allowed to adopt the book/books of their choice. Additional material will be provided by the instructor.