BASI MOLECOLARI DELLA VITA (INV112)

1. language

Italian.

2. course contents

Coordinator: Prof. ALESSANDRO LUPI
Anno di corso/Year Course: 1st
Semestre/Semester: 1st
UFC: 5
Modules and lecturers:
- BIOCHIMICA (INV05A) - 2 CFU - SSD BIO/10 - Prof. Alessandro Lupi
- BIOLOGIA (INV06A) - 1 CFU - SSD BIO/13 - Prof. Carmela Giampa'
- FISICA APPLICATA (INV08A) - 1 CFU - SSD FIS/07 - Prof. Luca Indovina
- GENETICA APPLICATA (INV07A) - 1 CFU - SSD MED/03 - Prof. Emanuela Abiusi

3. BIBLIOGRAPHY

Biochemistry Module
T. Bellini. Chimica medica e propedeutica biochimica con applicazioni cliniche Zanichelli 2017
G. Ricciotti, Fondamenti di Biochimica, Ed. Bovolenta, 1997.
Biology Module
Eldra P. Solomon, Linda R. Berg., Diana W. Martin) *Fondamenti di Biologia*, Edises
Hillis D. et al *Elementi di Biologia e Genetica*, Ed. Zanichelli.
Applied Physics Module
D. Scannicchio, Fisica Biomedica, EdiSES, 2013
Applied Genetics Module
Gelerther-Collins, Genetica Medica, Masson, 2000.
Neri G., Genuardi M. Genetica umana e medica (V edizione). Elsevier Masson 2024.
Students must have a textbook chosen from among the recommended ones or another textbook with the teacher's approval. The textbooks indicated for the courses may be supplemented by other teaching material that the teacher deems appropriate to highlight and include in their module to ensure adequate updating of the information.

4. LEARNING OBJECTIVES

The course aims to provide students with the skills necessary to understand the molecular basis of life and the biological processes underlying the functioning of the human body.
Knowledge and Understanding (Dublin 1) At the end of the course, students should demonstrate knowledge and understanding of the basics of biochemistry (compounds and metabolic reactions), biological processes, the principles of physics, and the mechanisms of

genetics.
Applied Knowledge and Understanding (Dublin 2) At the end of the course, students should demonstrate the ability to apply their acquired knowledge of biology and chemistry to interpret and explain biological phenomena. They should demonstrate the ability to use what they have learned to understand other disciplines and for practical application in analysis and research laboratories, recognizing that this knowledge is essential to understanding the application of specific techniques in the fields of biomedical diagnostics, therapy, and research.
Making Judgments (Dublin 3) At the end of the course, students should be able to independently discuss and critically analyze the cellular and molecular mechanisms underlying life processes. Independent judgment will be encouraged during lectures through discussions of relevant issues. During the test, students will also be assessed for their level of independent judgment.
Communication Skills (Dublin 4) By the end of the course, students should be able to present and explain their knowledge—even to non-experts—with logical rigor, proper language, and correct scientific terminology. Students should also be able to recognize and write the structural formulas of the main types of biomolecules. ***Learning Skills (Dublin 5)*** By the end of the course, students should be able to evaluate their knowledge and skills and, consequently, implement and/or update them independently by drawing on texts, scientific articles, and online platforms.

5. prerequisites

Knowledge of basic notions regarding Genetics (DNA), Biology (constitution of the cell), Chemistry, Mathematics (logarithms and graphs), and Physics is required.

6. TEACHING METHODS

The course is taught through lectures, which also include PowerPoint presentations (Dublin 1). During the lessons, students are actively involved through exercises and discussions (Dublin 2). Students thus begin to acquire independent interpretation of the importance of the mechanisms underlying the biology and chemistry of life (Dublin 3). They also acquire the specific terminology of a particular discipline and the ability to communicate with others (Dublin 4). Finally, students are encouraged to review the topic covered by studying the recommended texts and to express any doubts or curiosities in the following lesson. The disciplines will be taught in such a way as to lay the foundation and stimulate interest for further study (Dublin 5).

7. OTHER INFORMATION

Teachers are available to provide course information and clarify lessons by email or, for brief questions, at the end of the lessons.

8. METHODS FOR VERIFYING LEARNING AND FOR EVALUATION

A written exam with multiple-choice questions is required, supplemented, in special circumstances and at the candidate's request, by an oral examination on course topics. Students' preparation will be assessed based on their ability to describe biological and biochemical processes clearly and scientifically rigorously, and their ability to connect the various topics, demonstrating an understanding of the logic of metabolic routes. A weighted average is calculated, and students will receive a grade of 30/30 if they answer all questions correctly. The score of questions asked varies depending on their number (between 10 and 15 for each subject). Honors are awarded based on the unanimous decision of the examining commission. Partial grades from a test session will be retained only until the next one.

9. program

BIOCHEMISTRY

Atomic structure and the periodic table. Electronic configuration. Quantum numbers and orbitals. Isotopes. Radioactive decay. Chemical bonds. Electronegativity. Molecular structure. Molecules and chemical reactions. Water. Acid-base reactions. Oxidation number. Oxidation-reduction reactions. Thermodynamics: quantities and laws. Ideal gas law. Aqueous solutions of electrolytes. Definition of mole. Definitions of solution concentration. Colligative properties of solutions. Osmotic pressure and its biological effects. Physiological solutions. Definition of osmolarity. Acids, bases, and salts. Acid-base theories. Strong and weak acids. Dissociation constant, pKa. Ionic product of water and pH. Buffer solutions. Properties of the carbon atom. sp2 and sp3 hybridization. Nomenclature, chemical-physical characteristics, and reactivity of the main types of carbon compounds: hydrocarbons, alcohols, aldehydes and ketones, esters, carboxylic acids, nitrogenous compounds, aromatic compounds. Isomerism. Compounds of biological interest: amino acids, proteins, carbohydrates, lipids and phospholipids, nucleotides and nucleic acids. Enzymes: function and inhibition. Diseases caused by enzyme deficiencies. Main metabolic processes: glycolysis, Krebs cycle, beta-oxidation of fatty acids. Respiratory chain. Oxidative phosphorylation.

BIOLOGY

Discovery of the cells, basic properties and the two fundamentally different classes of cells. The structure and functions of biological molecules (carbohydrates, lipids, protein and nucleic acid). Differences between prokaryotic, eukaryotic cells and viruses. Cellular membrane (function and chemical composition, the movement of substances across cell membrane, diffusion and facilitated diffusion, active transport, exocytosis, endocytosis). Cellular organelles: nucleus, endoplasmic reticulum, lysosomes, golgi complex, mitochondrion and aerobic respiration). The cytoskeleton. The extracellular matrix and cell interactions. Cell division: cell cycle and its regulation. Mitosis and meiosis. Cell signaling pathways. Duplication and transcription of DNA. Translation of mRNA in prokaryotic and eukaryotic cells. Controlling gene expression.

APPLIED PHYSICS

Physical quantities and their units of measurement. Kinematics of the material point. Speed. Acceleration. Gravity acceleration. Circular motion. Dynamics and Newton's laws. Strength, weight, mass. Friction. Fundamental equations of the statics of rigid bodies. Levers. Elastic properties of materials. Elastic deformations, traction, compression, shear and torsion stresses. Momentum and conservation principle. Work, energy and its conservation. Mechanical power. Fluidostatics. Density and pressure. Stevino's law. Pascal's law. Archimedes' principle. Torricelli's experience and blood pressure gauges. Blood pressure measurement. Sphygmomanometer. Fluid dynamics.

Bernoulli equation. Torricelli's theorem. Venturi effect. Hydrodynamics of blood circulation. Surface tension. Capillarity. Diffusion. Fick's law. Membranes. Gases and solutions. Osmotic pressure. Osmosis processes in the biological field. Thermometric scales. Laws of perfect gases. Thermal capacity and specific heats. Phase changes, latent heat, heat propagation. Principles of thermodynamics. Electrostatics. Electrical properties of matter. Coulomb's law. Electric field and electric potential. Electric current and measuring instruments. Electrical resistance. Ohm's law. Operating principle of the electrocardiogram. Magnetic fields produced by electric currents. Electromagnetic radiation. Non-ionizing radiation. Ionizing radiation and interaction with biological matter.

APPLIED GENETICS

The human genome structure: genes and chromosomes.

Interpretation of the clinical significance of DNA variants: definition and classifications of genetic variants (variants, mutations, polymorphisms) and their role in human pathology: effects on mRNA (synonym, missense, nonsense, frameshift, affecting splicing sites); effects on protein function (gain of function, loss of function, silent); benign and pathogenetic variants; constitutional and somatic variants; mosaicism; inherited and *de novo* variants; homozygosity and heterozygosity.

Genetic basis of diseases: mendelian characters and hereditary diseases with mendelian transmission: autosomal dominant, autosomal recessive, X-linked; exceptions to mendelian transmission. Atypical mechanisms of inheritance: mitochondrial inheritance, microsatellite expansion diseases, imprinting defects.

Principles of cytogenetics: structure and number of human chromosomes; karyotype; mitosis and meiosis; crossing over; chromosomal anomalies and their classification.

Multifactorial diseases and genetic predisposition factors.

Role of genetic variants in the onset of tumors: notes on oncological genetics.

Invasive and non-invasive prenatal diagnosis; indication for carrying out pre- and post-natal genetic tests.