BASI MOLECOLARI DELLA VITA (INC112)

1. language

Italian.

2. course contents

Coordinator: Prof. ALESSANDRO LUPI
Year Course: 1st
Semester: 1st
UFC: 5
Modules and lecturers:
- BIOCHIMICA (INC05A) - 2 CFU - SSD BIO/10 - Prof. Alessandro Lupi
- BIOLOGIA (INC06A) - 1 CFU - SSD BIO/13 - Prof. Anna D'Altocolle
- FISICA APPLICATA (INC08A) - 1 CFU - SSD FIS/07 - Prof. Andrea Fidanzio
- GENETICA APPLICATA (INC07A) - 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
Elementi di Biologia e Genetica (Hillis D. et al., Ed. Zanichelli).
Applied Physics Module
Elementi di Fisica, V. Monaco, R. Sacchi. A. Solano, Ed. McGraw-Hill Education
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 short 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

The scientific process and introduction to biology. Cell theory. The characteristics of living organisms. Macromolecules of biological interest and their role in cell biology: organic compounds (carbohydrates, lipids, proteins, and nucleic acids). Cell organization: differences between prokaryotes and eukaryotes. Cell membranes: structure, function, and types of transport (diffusion, active transport, endocytosis, exocytosis, osmosis). Cytoplasmic organelles: structure and function (nucleus, nucleolus, smooth and rough endoplasmic reticulum, lysosome, Golgi apparatus, mitochondrion and cellular respiration, ribosomes). Extracellular matrix and cell adhesion. Cytoskeleton and cellular movement. DNA: structure and replication in prokaryotes and eukaryotes. Genetic code: Expression and translation of the genetic message: transcription and translation of RNA. Post-transcriptional RNA and post-translational protein maturation. The cell cycle, mitotic division, meiotic division, and its role in species evolution and genetic transmission.

APPLIED PHYSICS

Physical quantities; scalar and vector quantities; kinematics of a point; dynamics of a point. Statics: moment of force, static equilibrium, levers, mechanical advantage, levers in the human body, work and energy. Ideal fluids: fluids at rest, hydrostatic pressure, pressure measurement, Archimedes' principle; fluids in motion: laminar and turbulent flow, flow rate, continuity equation, blood flow, Bernulli's equation. Aneurysm and stenosis, TIA; Viscous fluids: friction, Poiseuille's law, blood

viscosity and hematocrit. Cardiac workload, vascular resistance, and pressure drop. Thermology: temperature, internal energy, heat, mechanical equivalent of a calorie, specific heat, heat transfer, first law of thermodynamics, and considerations of metabolism. Electrical phenomena: charge, Coulomb force, electric field, electric potential, current, Ohm's law, resistance in series and parallel, capacitance, defibrillator. Magnetic field, Lorentz force.

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.