

INTEGRATED TEACHING: BIOLOGY, APPLIED PHYSICS, BIOCHEMESTRY NUMBER OF CFU: 4 SSD: BIO/09 ; BIO/10 ; MED/03 ; BIO/13 RESPONSIBLE PROFESSOR: LAURA PACINI E-MAIL: Jaura.pacini@unicamillus.org

MODULE: BIOPHYSICS NUMBER OF CFU: 1 SSD: BIO/09 PROFESSOR: ELEONORA NICOLAI e-mail: <u>eleonora.nicolai@unicamillus.org</u> Office hours (by appointment) : Thursday from 3 pm to 4 pm

MODULE: BIOCHEMESTRY NUMBER OF CFU: 1 SSD: BIO/10 PROFESSOR: ELEONORA NICOLAI e-mail: <u>eleonora.nicolai@unicamillus.org</u> Office hours (by appointment) : Thursday from 3 pm to 4 pm

MODULE: MEDICAL GENETICS NUMBER OF CFU: 1 SSD: MED/03 PROFESSOR: MARIA ROSARIA D'APICE e-mail: mariarosaria.dapice@unicamillus.org Office hours (by appointment) : Thursday from 3 pm to 4 pm

MODULE: APPLIED BIOLOGY NUMBER OF CFU: 1 SSD: BIO/13 PROFESSOR: LAURA PACINI e-mail: <u>laura.pacini@unicamillus.org</u> Office hours (by appointment) : Thursday from 3 pm to 4 pm

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## PREREQUISITES

Knowledge and skills of basic Mathematics, Physics and Statistics at High School level, appropriate knowledge of the basic chemistry concepts including: chemical bonds, properties of solutions, acids, bases, buffers.

There are no prerequisites for the Biology module.

# **LEARNING OBJECTIVES**

The purpose of teaching is to provide students with the knowledge necessary for the performance of their future work. In particular, the comprehension of the physical principles underlying medical physics and the operation of medical instrumentation will be addressed.

By the end of the course, students will know the basic concepts of applying the Scientific Method to the study of biomedical phenomena (choice and measurement of parameters, evaluation of errors). They will be able to describe physical phenomena of complex systems using appropriate mathematical tools, they will know the scientific basis of medical procedures and the principles of operation of equipment commonly used for diagnostics and therapy.

They will acquire basic knowledge about the structure and function of biological macromolecules (carbohydrates, lipids, amino acids and proteins); basic knowledge about the main metabolic pathways and cycles with special emphasis on glucose, lipid and amino acid metabolism; knowledge about the morphological and physiological characteristics of the cell as a functional unit of living organisms. Another important objective will be the application of the experimental method as a means of understanding the biological mechanisms that regulate life and a tool for the study of pathological processes.

At the end of the course the student will also be able to distinguish the main classes of genetic, monogenic, chromosomal and multifactorial diseases and recognize their modes of transmission.

## **LEARNING OUTCOMES**

The specific learning outcomes of the program are coherent with the general provisions of the Bologna Process and the specific provisions of EC Directive 2005/36/EC. They lie within the European Qualifications Framework (Dublin Descriptors) as follows:

## **Knowledge and Understanding**

- Understand the experimental method and learn the use and transformation of measure units.
- Know and understand the proper terminology of physics.
- Know and understand the main physical principles and laws concerning electricity, vibration and waves, radiationheat and fluids.
- Apply these concepts to biological and physiological phenomena in living organisms.
- Identify and recognize the physical principles which govern the function of the specific human organs.
- Know basic information about the structure and function of major biological macromolecules.
- Know basic principles of enzymatic catalysis.
- Know the different metabolic pathways of eukaryotic cells.



- Know the role of different "fuels" in energy production.
- Know the biosynthetic pathways of some molecules of biochemical interest.
- Know what approaches and tools are used to study the cell.
- Describe bacteria and viruses.
- Know the differences between prokaryotic and eukaryotic cell.
- Know the structure and function of biological membranes.
- Describe cellular compartments and intracellular organelles.
- Describe physiology of the cell, the movement of molecules, passive transport, active transport, endocytosis (phagocytosis and pinocytosis) and exocytosis.
- Describe the nucleic acids. DNA and RNA. Transcription and translation. Regulation of gene expression.
- Describe the cell cycle.
- 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.
- Know and understand how to reconstruct family pedigrees and to calculate disease recurrence.
- Know and understand the major kinds of genetic testing and their proper use.

## Applying knowledge and understanding

- Apply the principles of physics to selected problems and a variable range of situations.
- Use the tools, methodologies, language and conventions of physics to test and communicate ideas and explanations.
- Adequately interpret the importance of alteration in biochemical processes alterations as the cause of various disease states.
- Use the acquired knowledge for an in-depth study of aspects related to his future professional activity.
- Ability to analyse family history to construct pedigrees.
- Ability to calculate disease recurrence risk.
- Use the acquired knowledge to understand the biological phenomena that regulate life and pathological processes.

## **Communication skills**

- Explaining arguments orally in an organized and coherent manner.
- Use of scientific language appropriate to and consistent with the topic of discussion.
- Communicate scientific and applied content clearly and unambiguously, using appropriate technical language.
- Describe the main patterns of inheritance and risk of recurrence and use correct genetic terminology.

#### Making judgements

- Recognize the importance of an in-depth knowledge of the topics consistent with a proper medical education.
- Identify the fundamental role of a proper theoretical knowledge of the topic in the clinical practice.



- Carry out assessments of the topics covered.
- Ability to synthesize and correlate the various topics.
- 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
- Make assessments, when related to the covered topics.

### **COURSE SYLLABUS**

### Syllabus BIOPHYSISCS

- Fundamental and derived physical quantities
- Dimensional equations
- Scientific Notation
- Orders of Greatness
- Scalar and vector quantities
- Vectors
- Mechanics
  - o Kinematics
  - o Uniform rectilinear motion
  - Rectilinear motion uniformly accelerated
  - Graphic representation of the motions
  - Uniform circular motion
- Dynamics
  - o Fundamental forces
  - Principles of dynamics: Newton's I, II, III law
  - Translational equilibrium
  - o Inertial and non-inertial reference systems
  - Inertial mass concept
  - Gravitational force
  - Strength weight
  - Normal force to the supporting surface
  - Tension of a rope
  - Frictional force
  - Centripetal Force / Centrifugal Force
  - o Electrostatic force
  - Elastic force
  - Work of a force
  - o Power
  - Kinetic and potential energy
  - Kinetic energy theorem
  - Potential energy theorem
  - Conservative and non-conservative forces
  - o Principle of conservation of mechanical energy



- $\circ \quad \text{Definition of yield} \\$
- Static
  - Moment of a force with respect to a point
  - Rotational balance
  - o Stable, unstable, indifferent equilibrium
  - o Simple machines: levers and pulleys
- CALORIMETRY
  - o Physical quantities that characterize a thermodynamic system: pressure, volume, temperature
  - o Thermometric scales
  - o Heat
  - Phase transitions
  - o Ideal gas law
  - o Thermal expansion
  - o Latent heat
  - o Heat transfer
- FLUIDS
  - o Hydrostatic
  - o Pressure
  - Pascal's principle
  - Stevino's law
  - Archimedes' principle
- Hydrodynamics
  - Law of continuity
  - o Bernoulli's theorem
  - Venturi effect
  - Poiseuille equation
- ELECTROSTATICS
  - o Coulomb's force
  - o Electric field
  - o Electric potential
  - o Electric current
  - Ohm's Laws
  - o Elementary electrical circuit: resistors in series and in parallel

## Syllabus BIOCHEMESTRY

Short summary of basic concepts of inorganic and organic chemistry - Chemical bonds, osmotic pressure, pH, buffers. The constituents of biological macromolecules: carbohydrates, lipids, purines, pyrimidines, nucleosides, nucleotides, amino acids. Proteins structure and function. Hemoproteins and gas transport (O2, CO2). Coenzymes and vitamins. Enzymes. Introduction to metabolism. Catabolism and anabolism. Glucose catabolism: glycolysis and the Kreb's cycle. Catabolism of fatty acids. The mitochondrion as the power plant of the cell: oxidative phosphorylation. Hormonal control of glucose metabolism. Insulin and glucagon: glycogenolysis, glycogen synthesis, gluconeogenesis and lipolysis. Fasting, diabetes and



ketogenesis. Biosynthesis of fatty acids and phospholipids. Cholesterol metabolism. Amino acid metabolism and urea cycle in brief.

# Syllabus MEDICAL GENETICS

- Basic Genetics: Definitions of Key Terms: gene, locus, allele, genotype, phenotype, haplotype, homozygous, heterozygous, haploid, diploid, dominance, recessivity, mutation, polymorphism.
- Principles of Genetic Transmission: Segregation in Human Pedigrees.
- Monogenic Inheritance Models: Autosomal inheritance, Autosomal recessive inheritance, X-linked inheritance
- Genetic Risk calculation and pedigrees
- Chromosomes: Structure and Analysis, Chromosomes Pathologies
- Genomic Imprinting
- X-chromosome inactivation
- Mitochondrial inheritance: mitochondrial DNA, pattern of inheritance
- Multifactorial inheritance: polymorphisms, susceptibility genes, gene-environment interaction, association studies
- Pharmacogenomics and Personalised Medicine
- Genetic tests and Counselling

# Syllabus APPLIED BIOLOGY

- Characteristics of living organisms, levels of organization and classification principles.
- Macromolecules structure, shape and function: carbohydrates, lipids, proteins and nucleic acids.
- The cell as the basic unit of life, Cell Theory. Prokaryotic and eukaryotic cells.
- Structure and function of the eukaryotic cell: plasma membrane, cytoplasm, ribosomes, smooth and rough endoplasmic reticulum, Golgi apparatus, lysosomes, peroxisomes, cytoskeleton.
- Relationship between energy conversion processes and cellular structures, mitochondria and chloroplasts (notes).
- Nucleus. Nuclear envelope, nucleoli, chromatin and chromosomes.
- Molecular bases of hereditary information. DNA structure and function.
- Gene expression: transcription and maturation of transcripts.
- Genetic code and translation. Main post-translational modifications and post-synthetic fate of proteins.
- Endomembranes and vesicular trafficking. Exocytosis and Endocytosis.
- Cell cycle, Mitosis and meiosis.

## COURSE STRUCTURE

The 4 modules are structured in 14 hours of frontal teaching each, divided into lectures of 2 or 4 hours, depending on the academic calendar. Frontal teaching includes theoretical lectures on the program topics for each module. The lecturers make use of educational tools such as presentations organized in powerpoint files with diagrams, illustrations and explanatory images.



In the Applied Biophysics module, a recovery of the mathematical concepts and skills that are indispensable prerequisites for a fruitful conduct of the Integrated Course will be carried out prior to the course.

The Biology module will make use of movies and animations to complement the description of the cellular processes analysed in class.

The Genetics module will include both theory lectures with power-point presentations and interactive lectures and exercises (either alone or in groups).

Attendance is mandatory. Attendance of at least 75 percent of the total scheduled hours for all lectures in the integrated course is required.

## **COURSE GRADE DETERMINATION**

Student learning will be assessed through a written test on topics related to Biology, Biophysics, Biochemistry and Medical Genetics.

The exam will be organized into 30 multiple-choice questions. Each question will consist of 4 or 5 answers of which only one is correct. Each correct answer is given a score of 1, each incorrect answer is given a score of 0 (there is no penalty for incorrect answers). If the written test results in a sufficient grade, the student is given the opportunity to improve the grade obtained by an optional oral test. The exam will cover the main topics of the teaching modules and will be considered passed if the student scores a final mark of 18/30.

The knowledge and ability to understand, the ability to apply knowledge and understanding, the autonomy of judgment and the communication skills of the student will weigh in the final score as follows 30%, 30%, 30% and 10%, respectively.

The evaluation criteria considered will be: acquired knowledge, independent judgment, communication skills and learning skills. The exams will be assessed according to the following criteria:

< 18 insufficient	The candidate possesses an inadequate knowledge of the topic, makes significant errors in applying theoretical concepts, and shows weak presentation skills.
18 - 20	The candidate possesses a barely adequate and only superficial knowledge of topic, limited presentation skills, and only an inconsistent ability to apply theoretical concepts.
21 – 23	The candidate possesses an adequate, but not in-depth, knowledge of the topic, a partial ability to apply theoretical concepts, and acceptable presentation skills.
24 – 26	The candidate possesses a fair knowledge of the topic, a reasonable ability to apply theoretical concepts correctly and present ideas clearly.
27 - 29	The candidate possesses an in-depth knowledge of the topic, a sound ability to apply theoretical concepts, good analytical skills, clear argumentative clarity and an ability to synthesize



**30 - 30L** The candidate possesses an in-depth knowledge of the topic, an outstanding ability to apply theoretical concepts, a high level of argumentative clarity, as well as excellent analytical skills, and a well-developed ability to synthesize and establish interdisciplinary connections.

### **OPTIONAL ACTIVITIES**

In addition to the teaching activity, the student will be given the opportunity to participate in seminars, research internships, department internships and monographic courses. The topics of the activities are not subject to examination. Acquisition of the hours allocated occurs only with a mandatory frequency of 100%.

### **READING MATERIALS**

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.

### **Reading materials for BIOPHYSISCS**

- Monaco, V., Sacchi, R., & Solano, A. M. (2007). Elementi di fisica.
- Scannicchio, D., Giroletti, E. (2015). Elementi di Fisica Biomedica. Edises.
- Bersani, F., Bettati, S., Biagi, P.F., Capozzi, V., Feroci, L., Lepore, M., Mita, D.G., Ortalli, I., Roberti, G., Viglino, P., Vitturi, A. (2009). Elementi di Fisica. Piccin.

#### **Reading materials for BIOCHEMESTRY**

• Ashok Kumar, J. (2011). Textbook of biochemestry. I K International Publishing House

## **Reading materials for MEDICAL GENETICS**

• Jorde, L.B., Carey, M.D., John, C. (2019). Medical Genetics. Elsevier Science Health Science.

## **Reading materials for APPLIED BIOLOGY**

- Curtis, H., Barnes, N.S., Schnek, M., Massarini, A. (2017). Elementi di biologia. Zanichelli.
- Sadava, D. E., Hillis, D. M., Heller, H. C., & Hacker, S. (2009). Elementi di biologia e genetica. Zanichelli
- Raven, P.H., Johnson, G.B., Mason, K.A., Losos, J.B., Singer, S.R. (2018). Elementi di biologia e genetica.
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