## BSc in Physiotherapy

**INTEGRADED COURSE TITLE:** **BIOLOGY, BIOCHEMISTRY AND APPLIED GENETICS**
**NUMBER OF ECTS CREDITS:** 6
**SSD: BIOS-07/A, BIOS-10/A, MEDS-01/A**
**MODULE CONVENOR:** PROF. MARIA PATRIZIA STOPPELLI
**E-MAIL:** mariapatrizia.stoppelli@unicamillus.org

MODULE: BIOCHEMISTRY
NUMBER OF ECTS CREDITS: 2
SSD: BIOS-07A
PROFESSOR: MARIA PATRIZIA STOPPELLI e- mail: mariapatrizia.stoppelli@unicamillus.org
Reception Time: by appointment

MODULE: MEDICAL GENETICS
NUMBER OF ECTS CREDITS: 2
SSD: MEDS-01/A
PROFESSOR: ANDREA LATINI e-mail **:** andrea.latini@unicamillus.org
Reception Time: by appointment

MODULE: APPLIED BIOLOGY
NUMBER OF ECTS CREDITS: 2
SSD: BIOS-10/A
PROFESSOR: BRUNI EMANUELE e-mail : emanuele.bruni@unicamillus.org
Reception Time: by appointment

### PREREQUISITES

Although there are no prerequisites and the basic concepts will be addressed in the first lessons of this integrated course dedicated to the "Additional Training Obligations" (OFA), a general knowledge of some basic notions of chemistry (atomic/molecular structure, valence, pH, molar concentration) and biology (properties of life, cellular theory, energy flows in ecosystems, concepts of meiosis/mitosis, DNA function) is required.

### LEARNING OBJECTIVES

The aim of this integrated course is to provide students with the knowledge of:

- the main classes of organic molecules and biological macromolecules, knowledge of the functioning of respiratory proteins and the working mechanism of enzymes;
- energy metabolism in humans and the role played by the main classes of biomolecules;
- the main metabolic pathways, including the main pathway of glucose catabolism;
- the morphological and functional organization of prokaryotic and eukaryotic cells, including the descriptive aspects, the basic biochemistry and cellular physiology required to understand the functions of the cell as the basic unit of living organisms;

- the constructive logic of the fundamental biological structures at the different levels of organization of living matter, the general unitary principles that govern the functioning of the different biological units, the learning of the experimental method and its applications to the study of biological phenomena.
- the inheritance of monogenic, chromosomal and multifactorial diseases and the analytic methodologies useful for the diagnosis of these pathologies.

At the end of the course the student will be able to know the main classes of macromolecules and their function, the mechanisms that regulate cellular activities, gene expression and the transmission of genetic heritage, to distinguish the main classes of genetic diseases and recognize the modes of transmission.

## LEARNING OUTCOMES

The expected learning outcomes are consistent with the general provisions of the Bologna Process and the specific provisions of Directive 2005/36/EC. They are found within the European Qualifications Framework (Dublin descriptors) as follows:

### Knowledge and Understanding

*At the end of this integrated course, the student will be able to:*

- *understand the structures of the most important biological macromolecules, framing the role of these molecules within the main metabolic processes that take place in the cell;*
- *know the principles of classification of living organisms;*
- *-describe the main characteristics and differences between prokaryotic and eukaryotic cells;*
- *describe cellular morphology;*
- *know the main cellular compartments and their function;*
- *know the general principles of cellular metabolism;*
- *know the differences between Mitosis and Meiosis;*
- *know the molecular basis of the transmission of hereditary characteristics*
- *know the correct genetic terminology, the main hereditary transmission models of monogenic, chromosomal and multifactorial diseases;*
- *know the main biological mechanisms that cause hereditary diseases;*
- *understand how to build family pedigrees and calculate the recurrence of the disease;*
- *understand the main types of genetic tests and their correct use.*

### Applying knowledge and understanding

The learning path of this course intends to provide the student with the knowledge and methodological tools for studying the biological bases of life, learning the experimental method and its applications to the study of fundamental biological phenomena. In particular, he will be able to:

- frame the role of molecules within the main metabolic processes occurring in living cells.
- assign metabolic processes to their respective subcellular compartments.

- -analyze family pedigrees together with clinical and molecular genetic data, useful for genetic counseling and calculating the risk of disease recurrence.

**Communication skills**
Students will be able to adequately describe a class of macromolecules, a biological phenomenon, the main inheritance models and the risk of recurrence, demonstrating that they have learned an appropriate scientific language for the purposes of correct, synthetic and rigorous communication.

**Making judgements**
Students will be able to independently develop the logical procedures and strategies for applying the experimental method, formulate a scientific hypothesis and correctly analyzing and interpreting experimental data, as well as using genetic tests for the molecular diagnosis of monogenic and chromosomal diseases or for the evaluation of genetic susceptibility to complex diseases.

**Learning skills**
The student will have acquired skills and learning methods suitable for deepening and improving their knowledge and skills in the subjects covered by integrated teaching, also through consultation of scientific literature.

**COURSE SYLLABUS**

**Syllabus Biochemistry**
OFAs: The structure of the atoms, isotopes, ions and their relationship to atomic number, mass number, and charge. 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: Aliphatic and aromatic hydrocarbons. Alcohols, aldehydes, ketones and carboxylic acids. Amines, esters, anhydrides and amides.
-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.
-Aminoacids, peptides and proteins. Peptide bond; primary, secondary, tertiary and quaternary structure of proteins.
Functions of proteins: Myoglobin and hemoglobin.
-Enzymes: characteristics and functioning; mechanisms of enzymatic inhibition.
-Nucleotides. Nucleic acids structure and function.
-Carbohydrate metabolism: glycogen, glycolysis and gluconeogenesis. Hormonal regulation of blood sugar.
-Amino acid metabolism: digestion of proteins and urea production.
-Bioenergetics: citric acid cycle, respiratory chain, electron transfer and ATP synthesis.
-Lipid metabolism: fatty acid catabolism, beta oxidation, ketogenesis.

**Syllabus 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: polymorphisms, susceptibility genes, gene-environment interaction, association studies
Pharmacogenomics and Personalised Medicine
Dynamic mutations and related disorders
Genetic tests and Counselling. Outlines

**Syllabus Applied Biology**
OFAs: Classification principles of living organisms. Cell theory. Characteristics of living cells. Classification and major structural differences between prokaryotic and eukaryotic cells. Structure and function of biomolecules. Cell metabolism, the use of energy by cells. Differences in asexual and sexual reproduction. Mitosis and Meiosis.

- Characteristics of living organisms: cell metabolism, reproduction. Cellular organization, types of macromolecules.
- Characteristics of living cells: cellular theory. Classification principles of living organisms.
- Prokaryotic and eukaryotic cell models: classification and major structural differences.
- Cell Chemistry: structure, shape and function of macromolecules. Proteins.
- Lipids. Structure, properties and function of plasma membrane.
- Internal organization of the cell: cellular compartments, cytoplasm and cytoplasmic organelles, ribosomes, smooth and rough endoplasmic reticulum, Golgi apparatus, lysosomes, peroxisomes, endomembranes and membrane traffic: endocytosis and exocytosis.
- Mitochondria and chloroplasts: glycolysis, fermentation, cellular respiration, photosynthesis (outline).
- Post-synthetic fate of proteins, protein sorting. Main post-translational modifications of the polypeptide chains.
- Cell cytoskeleton: microtubules, intermediate filaments and microfilaments. Cilia and flagella. Centrioles and centrosomes.
- Cell Nucleus: nuclear envelope, nucleoli, chromatin and chromosomes.
- Molecular basis of hereditary information: DNA replication. DNA repair and its correlation with human diseases.
- RNA structure and function: main types of RNAs and differences with respect to DNA in their molecular size, stability and biological functions. Transcription and RNA maturation.
- Genetic code and translation: The genetic code and its properties. Reading and interpretation of the genetic code. Protein synthesis.
- Cell cycle, mitosis and meiosis.

## COURSE STRUCTURE

The course is structured in 6 CFU with 60 hours of frontal lessons, including theoretical lessons with power-point presentations and exercises (both in groups and alone). Attendance is mandatory for, at least, 75% of the total hours of the integrated course.

## COURSE GRADE DETERMINATION

Student learning will be assessed through a written single test to be held at the end of the integrated course, in which the student will answer multiple choice questions on topics presented during the lessons. The written exam consists of 60 questions, 20 for each module. Correct answers will be given a score of 0.5.
The requirements for passing the exam are as follows:
- Correctly answer, at least, 50% of the questions (10/20) of each individual module
- Obtain a minimum overall score of 18/30. The student who obtain a score of 60/60 are admitted to compete for honors by taking an oral integration with the teachers.

## 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