Degree Programme in Medicine and Surgery

Academic Year 2026 / 2027

INTEGRATED COURSE (I.C.): Biology and Genetics
NUMBER OF CFU (ECTS credits): 8
SSD (Academic Discipline Code): BIOS-10/A, MEDS-01/A

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MODULE **Applied Biology**
NUMBER OF CFU (ECTS credits: **6**
SSD: **BIOS-10/A**
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MODULE **Medical Genetics**
NUMBER OF CFU (ECTS credits): **2**
SSD: **MEDS-01/A**

LECTURER:
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PREREQUISITE COURSES

Not required.

PREREQUISITES

Not applicable for the Biology module. For the Medical genetics module it is recommended that students have a solid background in biology, particularly in understanding gene structure, the DNA replication process and the concepts of mitosis and meiosis.

LEARNING OBJECTIVES

The integrated course in Biology and Genetics aims to provide students with a comprehensive understanding of the fundamental principles governing living systems, with particular emphasis on the cell as the structural and functional unit of life. Students will acquire knowledge of the molecular and cellular mechanisms that regulate cell structure, function, communication, and interactions, as well as the principles underlying the diversity of biological systems in relation to their structural, functional, and genetic characteristics.

The course also examines the mechanisms of gene expression and regulation in the context of cellular differentiation, organismal development, and biological evolution. Fundamental concepts of molecular biology and genetics will be covered, with particular emphasis on topics of medical relevance, including the cellular and molecular basis of human disease and the effects of drugs on cell structure and function.

The Medical Genetics module provides students with the essential principles underlying the inheritance of monogenic, chromosomal, and multifactorial disorders. By the end of the course, students will have acquired the fundamental knowledge and analytical approaches required for the diagnosis of genetic diseases, and will be able to distinguish the major classes of genetic disorders and understand their patterns of inheritance.

EXPECTED LEARNING OUTCOMES

The aim of the Biology and Genetics course is to provide students with a solid and integrated foundation in Biology and Medical Genetics, establishing the essential knowledge required to understand the physiological and pathological processes explored in subsequent biomedical courses.

DD 1: Knowledge and understanding

Understanding of the fundamentals of molecular and cellular biology. The student will acquire general and unitary principles governing the functioning and behavior of living organisms including the mechanisms involved in the transmission of hereditary traits.

At the end of this integrated teaching the student will be able to:

- Define a living organism
- Recognize the common characteristics of all living organisms.
- Describe the differences between prokaryotic and eukaryotic cells.
- Know the main cellular compartments and their function.

- Know the general principles of cellular metabolism.
- Know the molecular basis of transmission of hereditary traits.
- Understand the molecular basis of gene expression and its regulation.
- Know the differences between mitosis and meiosis.
- Know the basic mechanisms of cell differentiation.
- Recognize and describe the dysregulation consequences of the main processes operating in the cell and the molecular bases of the diseases.
- 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.
- Reconstruct family pedigrees and calculate disease recurrence.

DD 2: Applying knowledge and understanding

The overall aim of the integrated Biology and Genetics course is to introduce students to the scientific method and its application to the study of fundamental biological phenomena. Accordingly, the course seeks to foster the ability to make accurate, systematic, and well-documented observations, while developing the critical thinking and analytical skills required to formulate and evaluate evidence-based, testable hypotheses and generalizations. By the end of the course, students will be able to apply the scientific method to the investigation of biological processes and to understand and explain the molecular and cellular mechanisms underlying human disease. They will also acquire the knowledge and skills required to analyze family pedigrees and interpret clinical and molecular genetic data relevant to genetic counseling. In addition, students will be able to assess and calculate the recurrence risk of genetic disorders.

DD3: Making judgements

By the end of the course, students will have examined representative biological experiments and will be able to independently apply the logical approaches and strategies underlying the experimental method. They will be able to critically analyze and correctly interpret experimental data, integrate and correlate knowledge across different topics, and formulate evidence-based conclusions. In addition, students will be able to recognize the major patterns of inheritance and correctly classify genetic disorders as monogenic, chromosomal, or multifactorial.

DD4: Communication skills

Students will be able to accurately describe biological phenomena using appropriate scientific terminology and communicate biological and genetic concepts in a clear, precise, and rigorous manner. They will also be able to explain the major patterns of inheritance and estimate recurrence risk using appropriate genetic nomenclature and terminology.

DD5: Learning skills

Students will develop the learning strategies and independent study skills necessary to expand and update their knowledge in the fields of Biology and Medical Genetics. They will be able to critically consult and interpret the scientific literature as a resource for lifelong learning and for the continuous advancement of their scientific and professional knowledge.

SYLLABUS BIOLOGY

Characteristics of living organisms. Cell theory and the cell as the structural and functional unit of life.

The chemistry of life. Biological macromolecules: carbohydrates, lipids, proteins, and nucleic acids (structure and function).

Prokaryotic and eukaryotic cells. Classification and major structural differences. Organization of the eukaryotic cell. Cellular organelles and compartments: structure and function. Overview of viruses as intracellular parasites, including nucleic acid, capsid, and viral envelope.

Plasma membrane. The fluid mosaic model of the plasma membrane. Major functions of membrane proteins and their topological organization within the lipid bilayer. Transport of ions and small molecules across the plasma membrane: simple diffusion, facilitated diffusion, and active transport.

The nucleus. Nuclear envelope, nucleolus, chromatin organization and levels of chromatin condensation, chromosomes.

Molecular basis of genetic information. Structure and function of DNA. Identification of DNA as the genetic material. Molecular mechanisms of DNA replication, telomeres, and telomerase. Overview of DNA repair mechanisms.

Structure and function of RNA. Major RNA types in prokaryotic and eukaryotic cells. RNA transcription and RNA processing in eukaryotic cells, with particular emphasis on messenger RNA maturation. Roles of non-coding RNAs.

Protein synthesis. Ribosomes: structure, biological function, and differences between prokaryotic and eukaryotic ribosomes. Properties of the genetic code, general features of translation, and its biological significance.

Regulation of gene expression. Functional organization of the eukaryotic genome. Transcriptional regulation in prokaryotic and eukaryotic cells. Role of chromatin condensation. Histone code and DNA methylation (epigenetic modifications). Major post-transcriptional and post-translational regulatory mechanisms.

Compartmentalization of the eukaryotic cell. Cytoplasm and the endomembrane system: structure and function of the endoplasmic reticulum, Golgi apparatus, and lysosomes. Overview of peroxisomes.

Mitochondria. Structure and function of mitochondria as the cell's energy-producing organelles. Overview of glycolysis and cellular respiration. The endosymbiotic theory of mitochondrial origin.

Vesicular trafficking. Endocytosis (pinocytosis, phagocytosis, and receptor-mediated endocytosis) and constitutive and regulated exocytosis. Autophagy: macroautophagy, microautophagy, and chaperone-mediated autophagy.

Post-translational protein processing and trafficking. Overview of post-translational modifications and protein fate after synthesis. Role of the endoplasmic reticulum and Golgi apparatus in protein glycosylation. Protein targeting signals and mechanisms directing proteins to organelles and the secretory pathway.

Cytoskeleton, cell adhesion, and motility. Structure and function of intermediate filaments, microtubules, and actin filaments. Molecular motors. Cellular structures responsible for cell shape, polarity, and motility. Cell–cell and cell–extracellular matrix interactions. Cell adhesion molecules and the extracellular matrix.

Cell communication and signal transduction. Cell-to-cell communication in multicellular organisms. General principles of cell signaling, chemical signals, and receptor proteins.

Major signal transduction pathways. Nuclear receptors, G protein-coupled receptors, and enzyme-linked receptors. Second messengers, protein kinases, and molecular switches. Biological basis of membrane excitability: neurons and the action potential.

Mitosis and meiosis. The cell cycle, its phases, and regulation of cell cycle progression through the interaction of intracellular mechanisms and extracellular signals. Role of cyclin-dependent kinases (CDKs). Chromosome dynamics during mitosis and meiosis, and the major differences between these processes. Genetic consequences of meiosis and its importance as a source of genetic variation. Molecular mechanisms of genetic recombination. Concepts of haploidy and diploidy. Homologous chromosomes. Characteristics of sexual and asexual reproduction.

Mechanisms of cell death. Basic concepts of apoptosis, necrosis, and autophagy.

Molecular basis of cancer. Molecular mechanisms of cellular transformation. Hallmarks of cancer cells. Genetic and epigenetic alterations underlying tumor development. Tumor suppressor genes and proto-oncogenes.

SYLLABUS GENETICS

Basic Genetics: Definitions of Key Terms
Gene, locus, allele, genotype, phenotype, homozygous, heterozygous, haploid, diploid, dominance, recessivity, codominance, mutation, and polymorphism.

Principles of Genetic Transmission: Mendel's genetic hypotheses; monohybrid and dihybrid crosses; segregation in human pedigrees; genetics of blood groups.

Monogenic Inheritance Models: Autosomal inheritance, autosomal recessive inheritance, and X-linked inheritance.

Genetic Risk Calculation and Pedigree Analysis

Population Genetics: Hardy–Weinberg equilibrium and its theoretical implications for understanding the mechanisms of biological evolution.

Chromosomes: Structure and Analysis.
Chromosome structure and analysis; chromosomal abnormalities and genetic disorders.

X-Chromosome Inactivation.

Mitochondrial Inheritance.

Multifactorial Inheritance and Common Diseases: Polymorphisms, susceptibility genes, gene–environment interactions, and association studies.

COURSE STRUCTURE

The course is divided into classroom lectures, of which 60 hours dedicated to Biology and 20 hours dedicated to Medical Genetics. The instructors use various educational tools including organized PowerPoint presentations with explanatory diagrams, illustrations and images which are taken from micrographies to describe the various cellular structures. Videos and animations will be employed to complement the topics discussed in class. Interactive lessons with in-class exercises (both individual and group-based) are planned for the Genetic module. Intermediate tests may be scheduled for both the modules. The attendance at lectures is mandatory with a minimum requirement of at least 67% attendance of the total hours scheduled for all integrated course lectures.

COURSE GRADE DETERMINATION

The examination consists of two parts: a written test and an oral examination. The written test consists of 60 multiple-choice questions, 30 in Biology and 30 in Genetics, with only one correct answer, covering topics addressed during the course. Students must pass the written test in order to be admitted to the Biology oral examination. To be admitted to the oral examination, students must obtain a minimum score of 18/30 in the written test.

During the Biology oral examination, students are given the opportunity to demonstrate their knowledge by discussing the topics covered in the course and by reasoning through biology-related issues, demonstrating their ability to make connections between different concepts and to communicate using appropriate scientific terminology.

For students with certified disabilities or specific learning disorders, the appropriate compensatory measures and examination procedures established by the competent University offices are available.

Hence, the whole examination will be evaluated as it follows:

Fail	Significant gaps and/or inaccuracies in knowledge and understanding of the topics; limited analytical and synthesis skills; argumentation absent or frequently incorrect.
18–20	Barely sufficient knowledge and understanding of the topics, with possible imperfections; sufficient analytical, synthesis, and judgement skills.
21–23	Routine knowledge of the topics; correct analytical and synthesis skills with coherent logical argumentation.
24–26	Fair knowledge and understanding of the topics; good analytical and synthesis skills; rigorously expressed argumentation.
27–29	Complete knowledge of the topics; considerable analytical and synthesis skills; good independent judgement.

30–30L	Excellent level of knowledge and understanding. Considerable analytical, synthesis, and independent judgement skills. Original argumentation expressed with full command of specialized terminology.

OPTIONAL ACTIVITIES

Students may voluntarily participate in practical Biology sessions, during which they will have the opportunity to use a light microscope. In addition, students may attend the office hours offered by the instructors of the integrated course. Office hours are available by appointment, which should be arranged via email.

READING MATERIALS

The recommended textbooks should be considered as reference teaching material. However, students may choose the text(s) they prefer or deem most suitable for their learning needs. Additional material will be provided by the lecturer.

Texts MODULE 1

- "Essential Cell Biology", Bruce Alberts et al. V ed., WW Norton & Co.

For further information you can use the following text: "Molecular Biology of the Cell", Bruce Alberts et al., VII ed., WW Norton & Co.

Texts MODULE 2

- "Fundamentals of medical genetics", Dallapiccola Bruno, Novelli Giuseppe. Edited by Edizioni Scientifiche Falco;

or

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