Syllabus Master's Degree Course in Medicine and Surgery

THE CELL: MOLECULES AND PROCESSES 1

First year, first semester (5 academic credits [CFU])

Teachers

Subject	Academic credits (CFU)	Lecturer
Molecular biology 1	1	DE PINTO Vito
Applied biology 1	2	BARBAGALLO Davide
Medical genetics 1	2	ROMANO Corrado

Learning outcomes

Subject	Learning outcomes
Molecular biology 1	By the end of the course, students are expected to: • Have the basis for the understanding of the physical, chemical and biological contexts in which molecules, reactions and metabolic pathways are framed. • Highlight the relationships between structure and function of the main classes of macromolecules. • Understand the regulation of molecular processes at cellular level. • Develop an interest in and be introduced to experimental methods. At the end of the course the student will understand the structure-function relationships of the main biological molecules, the biochemical mechanisms essential for a correct metabolic function and the consequences of their alterations.
Applied biology 1	By the end of the course, students are expected to: • have a knowledge and understanding of general biology in terms of (i) application of the scientific method to solve biological questions; (ii) evolution of biological entities (from viral to prokaryotic and eukaryotic cellular organizations); (iii) main differences between eukaryotic and prokaryotic cellular organizations with particular emphasis on eukaryotic cells; • use a technical language related to biological issues during their presentation; • be proficient in interconnecting biological structures and functions;

	• be able to autonomously revise the theory acquired during the course in terms of evolution and to translate their knowledge to modern medical research and practice.
Medical genetics 1	By the end of the course, students are expected to: • Understand the meaning of medical genetics in health and disease. • Understand how and why genetic variation is crucial in health and disease. • Be proficient in analyzing Inheritance patterns, genetic penetrance, and phenotype variability in mendelian disorders. • Understand how zygosity may impact in health and disease.

Prerequisites

Subject	Prerequisites
Molecular biology 1	Basic knowledge of biochemistry and biology.
Applied biology 1	Basic knowledge of Chemistry and Physics.
Medical genetics 1	Basic knowledge of Applied and Molecular Biology.

Course contents

Subject	Course contents
Molecular biology 1	• THE NUCLEIC ACIDS: PRIMARY STRUCTURE. • DNA as the ideal molecule for the perpetuation of genetic information - Changes in DNA sequence can have consequences: mutations. • NUCLEIC ACIDS: SECONDARY STRUCTURE • Denaturation of DNA. Hybridisation and annealing - Secondary structure of ssNA - Supercoiling or twisting/relaxation of DNA - Topoisomerases • THE RNA MOLECULES • REPLICATION: - Replicon - origin - replication fork – DNA polymerases - Primasome and Replisome - Termination - - Telomeres and telomerase - Regulation of replication • REPAIR, RECOMBINATION AND REARRANGEMENT IN DNA Repair systems • PROTEIN SYNTHESIS - Ribosomes - Stages of protein synthesis: initiation / elongation / termination - differences between Bacteria and Eukaryotes - inhibitors of protein synthesis • GENETIC CODE - Central dogma of biology and its modifications; relationships between gene, mRNA, proteins - how many tRNAs exist? - tRNA-aminoacyl synthetase • TRANSCRIPTION - Components of the transcriptional unit: promoter-site initiator-transcriber-terminator - Importance of transcription in the regulation of gene expression • TRANSCRIPTION IN PROKARYOTS: RNA polymerases - Promoter and recognition modes – Regulatory genes and structural genes - Operons and their regulation

Applied biology 1	• The origin of life and cell theory
	  ○ The scientific method
	  ○ The discovery and advances of microscopy
	  ○ The prebiotic word (RNA word)
	  ○ The theory of evolution by natural selection

	○ Differences between homology and analogy ○ The emerging properties ○ The relationship between structure and function • The emergence of modern cell biology ○ The advent of cell biology ○ The advent of molecular biology ○ The critical importance of technology and use of model organisms • The chemistry of the cell ○ The main chemical elements of the cells ○ Water and its properties ○ Description of the main functional chemical groups with a “biological” meaning • The main classes of biomolecules and their importance in biology ○ Carbohydrates, Lipids, Proteins, Nucleic Acids • Viruses, viroids and prions ○ Prokaryotic viruses (hint of their classification and life cycle) ○ Eukaryotic viruses (hint of their classification and life cycle) • Cells and organelles ○ Structure and function of prokaryotic cells (Bacteria and Archaea): plasma membrane, cell wall, nucleoid ○ Structure and function of eukaryotic cells: the concept of “compartmentalization”, plasma membrane, nucleus, nucleolus, nucleoplasm, endoplasmic reticulum, ribosomes, mitochondria (the endosymbiotic theory), Golgi complex, lysosomes, peroxisomes, cytoskeleton (microfilaments, microtubules, intermediate filaments) • Chromatin and chromosomes • Cell membrane: structure, function, and chemistry ○ Cell membrane as a permeability barrier ○ Glucidic, lipidic and protein components of the cell membrane – the biological importance of asymmetric structure of cell membranes ○ The involvement of cell membrane in inflammation • Transport across membranes ○ Simple diffusion and osmosis ○ Facilitated diffusion ○ Primary and secondary active transport
Medical genetics 1	• The meaning of medical genetics in health and disease • Nuclear and Mitochondrial Genomes: from the cell to health and disease • Genome variability in health and disease • The wide range of genetic variants: 1) Copy Number Variant, 2) Sequence Variant, 3) Polynucleotide Repeat Expansion, 4) Imprinting defect • The genotype • The phenotype • Phenotype-first and Genotype-first approaches • Penetrance • Expressivity • Alleles and Genetic Loci • Ploidy: Haploid, Diploid and Polyploid sets • Zygosity: 1) Homozygosity, 2) Heterozygosity, 3) Hemizygosity, 4) Wildtype Homozygosity, 5) Mutated Homozygosity, 6) Simple Heterozygosity, 7) Compound Heterozygosity

Assessment methods

Subject	Assessment methods
Molecular biology 1	The final assessment of acquired knowledge is conducted by a written or oral examination. The grade is expressed on a scale of thirty, up to a maximum of 30/30 cum laude (with honors). The final grade is determined by the weighted average of the scores obtained in the course subjects. The written examination will consist of at least 30 questions with multiple choice answers. The oral examination will consist of an interview during which questions will cover at least three different topics from the course curriculum. The assessments aim to evaluate: i) the level of knowledge in the disciplines; ii) the ability to apply this knowledge to solve specific problems related to the disciplines (autonomous problem-solving); iii) clarity of expression; iv) proficiency in medical-scientific language. The assessment of learning can also be conducted remotely if the conditions necessitate it. For the assignment of the final grade, the following parameters will be considered: • Score 29-30 with honors: The student demonstrates an in-depth knowledge of the topics, promptly and correctly integrates and critically analyzes presented situations, independently solving even highly complex problems. They possess excellent communication skills and command medical-scientific language proficiently. • Score 26-28: The student has a good understanding of the topics, is able to integrate and critically and logically analyze presented situations, can fairly independently solve complex problems, and presents topics clearly using appropriate medical-scientific language. • Score 22-25: The student has a fair understanding of the topics, although it may be limited to the main areas. They can integrate and critically analyze presented situations, although not always in a linear fashion, and present topics fairly clearly with moderate language proficiency. • Score 18-21: The student has minimal knowledge of the topics, possesses modest ability to integrate and critically analyze presented situations, and presents topics sufficiently clearly, although their language proficiency may be underdeveloped. • Exam not passed: The student lacks the minimum required knowledge of the core content of the course. Their ability to use specific language is minimal or nonexistent, and they are unable to independently apply acquired knowledge.
Applied biology 1	
Medical genetics 1	

Examples of common questions and/or exercises

Subject	Examples of common questions and/or exercises
Molecular biology 1	• The alternative DNA conformations • Operon lac regulation • Genetic code degeneracy • Protein synthesis phases

Applied biology 1	• The principles of cell theory • Describe the main differences between eukaryotic and prokaryotic cell organizations • Describe the structure and function of plasma membrane • Describe the structure and function of cytoskeleton • Describe how does primary active transport across plasma membrane happen • What is chromatin • Describe how does Na+/K+ pump work
Medical genetics 1	• What's Medical Genetics • How Nuclear and Mitochondrial Genomes lead to a different genetics • The wide range of genetic variants • Genotype • Phenotype • Imprinting • Penetrance • Expressivity • Alleles and Zygosity

Reference texts

Subject	Textbooks
Molecular biology 1	• Zlatanova & K.E. vanHolde Molecular Biology. Structure and dynamics of Genomes and Proteomes, 1st edition, 2016, Garland Sciences, ISBN: 9780815345046 • James D. Watson et al, Molecular Biology of the Gene, 7th edition, 2014, Pearson, ISBN: 9780321762436. Any additional educational material (slides, videos, handouts, etc.) will be distributed or indicated during the lessons.
Applied biology 1	• Radin and Lodolce. Becker's world of the cell. Tenth edition, 2022, Global Edition • Alberts, Hopkin, Johnson, Morgan, Raff, Roberts, Walter. Essential cell biology. Fifth edition, 2019, Norton Any additional educational material (slides, videos, handouts, etc.) will be distributed or indicated during the lessons.
Medical genetics 1	• Strachan and Lucassen. Genetis and Genomics in Medicine. Second Edition, 2023, CRC presso, Taylor and Francis Group. • Pyeritz, Korf, and Grody. Emery and Rimoin's Principles and Practice of Medical Genetics and Genomics, 7th Edition, 2019, Elsevier. • Jorde, Carey, and Bamshad. Medical Genetics, 6th Edition, 2020, Elsevier. Any additional educational material (slides, videos, handouts, etc.) will be distributed or indicated during the lessons.

Course format

Subject	Textbooks
Molecular biology 1	The teaching will primarily be conducted through in-person lectures with a blend of theory and practical exercises. In the event that teaching is delivered in a blended or remote mode, necessary adjustments may be introduced compared to what has been previously stated, in order to adhere to the planned program as outlined in the Syllabus.
Applied biology 1	
Medical genetics 1	

Attendance

Subject	Textbooks
Molecular biology 1	Mandatory attendance.
Applied biology 1	
Medical genetics 1	

Course schedule

Subject	Textbooks
Molecular biology 1	• Structure of DNA and RNA (Watson chapters 4, 5) • Structure of Genes (Zlatanova chapter 7) • DNA-Protein Interactions (Zlatanova chapter 6) • Replication of DNA (Watson chapter 9) • Mutability and Repair of DNA (Watson chapter 10) (Zlatanova chap. 22) • Translation (Watson chapter 15) • Genetic Code (Watson chap. 16) (Zlatanova chapter 7) • Transcription (Watson chapters 13) • Transcriptional Regulation (Watson chapters 18)
Applied biology 1	• The origin of life and cell theory; The Emergence of Modern Cell Biology (Radin and Lodolce, chapter 1) • The chemistry of the cell (Radin and Lodolce, chapter 2) • The main classes of biomolecules and their importance in biology (Radin and Lodolce, chapter 3; Alberts et al., chapters 2, 4, 5) • Viruses, viroids and prions (Radin and Lodolce, chapter 4) • Cells and organelles (Radin and Lodolce, chapters 4, 12, 13, 16; Alberts et al., chapter 17) • Chromatin and chromosomes (Alberts et al., chapter 5) • Cell membrane: structure, function, and chemistry (Radin and Lodolce, chapter 7) Transport across membranes (Radin and Lodolce, chapter 8)
Medical genetics 1	• Fundamentals of DNA, chromosomes and cells (Strachan and Lucassen, chapter 1) • Fundamentals of human genome organization (Strachan and Lucassen, chapter 2) • Principles of genetic variation (Strachan and Lucassen, chapter 4) • Single-gene disorders: inheritance patterns, phenotype variability, and allele frequencies (Strachan and Lucassen, chapter 5) • Principles of gene regulation and epigenetics (Strachan and Lucassen, chapter 6)