Syllabus Master's Degree Course in Medicine and Surgery

THE CELL: MOLECULES AND PROCESSES

First year, first semester (7 academic credits [CFU])

Teachers

Subject	Academic credits (CFU)	Lecturer
Molecular biology I	2	DE PINTO Vito
Applied biology I	3	BARBAGALLO Davide
Medical genetics I	2	ROMANO Corrado

Learning outcomes

Subject	Learning outcomes
Molecular biology I	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 I	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 I	By the end of the course, students are expected to: • Demonstrate a comprehensive understanding of the fundamentals of DNA, chromosomes, and cells. • Possess a deep knowledge of gene structure, gene expression, and human genome organization. • Exhibit a solid grasp of the principles underlying core genetic Technologies. • Understand the principles of genetic variation. • Be proficient in analyzing Inheritance patterns, phenotype variability, and allele frequencies in single-gene disorders. • Comprehend the principles of gene regulation and epigenetics.

Prerequisites

Subject	Prerequisites
Molecular biology I	Basic knowledge of biochemistry and biology.
Applied biology I	Basic knowledge of chemistry and physics.
Medical genetics I	Attainment of the educational objectives set by prerequisite courses.

Course contents

Subject	Course contents
Molecular biology I	• THE NUCLEIC ACIDS: PRIMARY STRUCTURE. • DNA as the ideal molecule for the perpetuation of genetic information - Changes in DNA sequence can have consequences: mutations. • THE EUKARYOTIC AND PROKARYOTIC GENOMES • INTERRUPTED GENES • NUCLEIC ACIDS: SECONDARY STRUCTURE • Denaturation of DNA. Hybridisation and annealing - Secondary structure of ssNA - hairpins - Alternative DNA conformations - Interaction with proteins - protein-DNA recognition language - protein DNA-binding domains - Supercoiling or twisting/relaxation of DNA -Topoisomerases • THE RNA MOLECULES: Central dogma of biology and its modifications - general concepts: gene expression; relationships between gene, mRNA, proteins • REPLICATION: an event related to cell duplication - Replicon - origin - replication fork – DNA polymerases - Primasome and Replisome - Termination - Termination of linear replicons and their initiation - Telomeres and telomerase - Regulation of replication • REPAIR, RECOMBINATION AND REARRANGEMENT IN DNA Repair systems • RECOMBINATION - homologous recombination - site-specific recombination • TRANSPOSONS: their action on evolution - transposons IS and Tn - Mechanisms of transposition

	• RETROSEQUENCES - Retrotransposons - Retroviruses - reverse transcriptase - unprocessed pseudogenes, Alu seq. and repeated sequences in genomes • PROTEIN SYNTHESIS - Ribosomes - Stages of protein synthesis: initiation / elongation / termination - differences between Bacteria and Eukaryotes - inhibitors of protein synthesis • GENETIC CODE - how many tRNAs exist? - tRNA-aminoacyl synthetase • TRANSCRIPTION - mechanism and general concepts - Components of the transcriptional unit: promoter-site initiator-transcriber-terminator - Importance of transcription in the regulation of gene expression - General steps of transcription • TRANSCRIPTION IN PROKARYOTS: RNA polymerases - Promoter and recognition modes – Regulatory genes and structural genes - Types of Operons and their regulation • TRANSCRIPTION IN EUKARYOTS: RNA polymerase of three types - Transcription factors: General factors, Upstream factors or enhancers, Inducible factors (response elements) - Promoters – Enhancer, Silencer, Mediator. • RNA PROCESSING - Eukaryotic mRNA maturation – 5'cap - polyA tail - base modifications in tRNAs - Alternative splicing - RNA editing • TRANSCRIPTION REGULATION IN EUKARYOTS - TRANSCRIPTION FACTORS - the response element - Other types of transcriptional regulation: - interaction with chromatin - long-distance regulation: hypersensitive sites, isolators, LCR (locus control region), - Methylation: CpG islands

Applied biology I	• 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)
	• Hints of bioenergetics and enzymes
	• 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 I	• Fundamentals of DNA, chromosomes and cells
	  ○ Structure and function of chromosomes
	  ○ DNA and chromosomes in cell division and the cell cycle
	• Fundamentals of human genome organization
	  ○ RNA genes and noncoding RNA
	  ○ Details and meaning of the human genome
	  ○ Electronic resources for the interrogation of the human genome sequence and gene products
	  ○ The organization and evolution of the human genome
	• Principles of genetic variation
	  ○ DNA sequence variation origins and DNA repair
	  ○ Population genomics and the scale of human genetic variation
	  ○ Functional genetic variation and protein polymorphism
	  ○ Extraordinary genetic variation in the immune system
	• Single-gene disorders: inheritance patterns, phenotype variability, and allele frequencies
	  ○ Terminology, electronic resources, and pedigrees
	  ○ Mendelian and mitochondrial DNA inheritance patterns
	  ○ Uncertainty, heterogeneity, and variable expression of mendelian phenotypes
	  ○ Allele frequencies in populations
	• Principles of gene regulation and epigenetics
	  ○ Genetic regulation of gene expression
	  ○ Chromatin modification and epigenetic factors in gene regulation
	  ○ Abnormal epigenetic regulation in mendelian and imprinting disorders

Assessment methods

Subject	Assessment methods
Molecular biology I	Since it is an annual course, the exam will be held in the second semester along with the second module. The final assessment of acquired knowledge is conducted by an 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 oral examination consists 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
Applied biology I	
Medical genetics I	

	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.

Examples of common questions and/or exercises

Subject	Examples of common questions and/or exercises
Molecular biology I	• The alternative DNA conformations • Operon lac regulation • Mechanism of DNA modifying enzymes • Genetic code degeneracy • Protein synthesis phases
Applied biology I	• The principles of cell theory • Describe the main differences between eukaryotic and prokaryotic cell organizations • Describe how does a microscope work • 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 I	• The flow of genetic information within the cell • Autosomal Dominant Inheritance • Autosomal Recessive Inheritance • X-Linked Inheritance • Mitochondrial Inheritance

	• Dynamic Mutations • Imprinting • Penetrance • Expressivity • Mutations and Genetic Variants • Alleles and Zygosity

Reference texts

Subject	Textbooks
Molecular biology I	• 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 I	• 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 I	• 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 I	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 I	
Medical genetics I	

Attendance

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

Course schedule

Subject	Textbooks
Molecular biology I	• 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) • Recombination (Zlatanova chapter 21) • Translation (Watson chapter 15) • Genetic Code (Watson chap. 16) (Zlatanova chapter 7) • Transcription (Watson chapters 13, 14) • Transcriptional Regulation (Watson chapters 18, 19)
Applied biology I	• 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) • Hints of bioenergetics and enzymes (Alberts et al., chapter 3) • 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 I	• 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)