Syllabus Master's Degree Course in Medicine and Surgery

THE CELL: MOLECULES AND PROCESSES

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

Teachers

Subject	Academic credits (CFU)	Lecturer
Molecular biology II	1	IRACI Nunzio
Applied biology II	2	RAGUSA Marco
Medical genetics II	2	ROMANO Corrado

Learning outcomes

Subject	Learning outcomes
Molecular biology 2	By the end of the course, students are expected to: • Integrate the knowledge of prokaryotic processes with the advanced concepts of eukaryotic cells. • Understand the basic concepts and terminology of the main techniques of molecular biology. • Describe the applicability of the main techniques of molecular biology. • Discuss the connections between the molecular approaches and the notions described in the whole course.
Applied biology 2	The course aims to provide information to understand the general and advanced principles on which life is based. The main educational purposes of the course are the following: • knowledge of the basic mechanisms of transmission of signals among cells • understanding of the principles of differentiation, replication and death of the cells • knowledge of epigenetic bases and the role of non-coding RNAs •
Medical genetics 2	By the end of the course, students are expected to: • Understand the meaning of a sporadic or inherited phenotype • Identify and manage all kinds of genetic inheritance • Understand the relationship between a genetic variant and its effect on the phenotype • Analyze and discuss the principles and key concepts of cancer genetics and genomics, including the genetic basis of various types of cancer

	• Be proficient on the rules, the meaning and the workflow of a genetic clinic • Apply genetic approaches to the development and treatment of diseases, including the use of genetic therapies and targeted interventions.

Prerequisites

Subject	Prerequisites
Molecular biology 2	Basic notions of Biochemistry and Molecular Biology.
Applied biology 2	Basic notions of Biology and Genetics.
Medical genetics 2	Medical Genetics I.

Course contents

Subject	Course contents
Molecular biology 2	• 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 - 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). • Manipulation of nucleic acids: electrophoresis and nucleic acid hybridization. • Polymerase chain reaction (PCR), reverse transcription PCR (RT-PCR) and quantitative PCR (qPCR). • DNA sequencing and Next Generation Sequencing (NGS). • Gene expression and study of proteins: chromatography; western blot (WB); enzyme-linked immunosorbent assay (ELISA).
Applied biology 2	Epigenetics • Chromatin modification • DNA methylation • Non-coding RNAs and gene expression regulation Signal Transduction Mechanisms • Chemical signals and cellular receptors • G-protein coupled receptors • Enzyme-coupled receptors • Synaptic transmission • Hormones and other long-range signals The Cell Cycle and Mitosis • Nuclear and cell division • Regulation of cell cycle • Growth factors and cell proliferation • Sexual Reproduction and Meiosis Apoptosis

	• Death signals and survival factors • Apoptotic pathways • Stem cells
Medical genetics 2	• Inherited and Sporadic Phenotype • Mendelian Inheritance: 1) Autosomal Dominant, 2) Autosomal Recessive, 3) X-linked Recessive, 4) X-linked Dominant • Mitochondrial Inheritance • Dynamic Mutations • Gonadal and Somatic Mosaicism • The variants and their effect on the phenotype: 1) Synonymous variant, 2) Nonsynonymous variant, 3) Missense variant, 4) In-frame CNV, 5) Frameshift CNV, 6) Splicing variant • The concept of multifactorial inheritance • The role of epigenetics in modulating development, health, and disease • Cancer Genetics and Genomics: from mutated genes to progressive genomic instability • The genetic clinic and its multifaceted role: 1) Disease ascertainment and follow, 2) Risk assessment for patients and family members, 3) Genetic counseling before conception, during preimplantation, during prenatal life, and after birth, 4) Presymptomatic genetic counseling, 5) The correct prevision of a genetic test Genetic approaches to the treatment of diseases • overview of treating genetic diseases and the genetic treatment of the diseases • small molecule drugs and therapeutic proteins • principles of gene and cell therapy gene therapy for inherited disorders

Assessment methods

Subject	Assessment methods
Molecular biology 2	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
Applied biology 2	
Medical genetics 2	

	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 2	• Differences between prokaryotic and eukaryotic transcription • Main steps in RNA maturation • The mechanism of PCR • Strategies of DNA sequencing
Applied biology 2	• Biological meaning of apoptosis • The role of non-coding RNAs in post-transcriptional regulation • The role of G-protein coupled receptors • Control of cell cycle
Medical genetics 2	• Autosomal Dominant Inheritance • Autosomal Recessive Inheritance • X-linked Inheritance • The effect of variants on the phenotype • The impact of environment and epigenetics in the phenotype • Cancer genetics • Genetic services • Genetic treatment of the diseases

Reference texts

Subject	Textbooks
Molecular biology 2	• J. 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 2	• Becker's World of the Cell, Global Edition, Hardin and Lodolce, tenth edition. Pearson. • Any additional educational material (slides, videos, handouts, etc.) will be distributed or indicated during the lessons.
Medical genetics 2	• 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 2	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 2	
Medical genetics 2	

Attendance

Subject	Textbooks
Molecular biology 2	Mandatory attendance.
Applied biology 2	
Medical genetics 2	

Course schedule

Subject	Textbooks
Molecular biology 2	• Transcription and RNA maturation (Watson chapters 13, 14) • Transcriptional Regulation (Watson chapters 19, 20) • The different techniques are distributed along the different chapters of the books and will be further implemented with didactic material provided by the teacher.
Applied biology 2	• Epigenetics: Becker's World of the Cell (chapter 20) + didactic material provided by the teacher • Signal Transduction Mechanisms: Becker's World of the Cell (chapter 22, 23) • The Cell Cycle and Mitosis: Becker's World of the Cell (chapter 24) • Sexual Reproduction and Meiosis: Becker's World of the Cell (chapter 25) • Apoptosis: Becker's World of the Cell (chapter 25) + didactic material provided by the teacher • Stem cells: didactic material provided by the teacher
Medical genetics 2	• The pathogenic role of genetic variation (Strachan and Lucassen, chapter 7) • The identification of disease genes and genetic susceptibility to complex diseases (Strachan and Lucassen, chapter 8) • Genetic approaches to the treatment of diseases (Strachan and Lucassen, chapter 9) • Cancer genetics and genomics (Strachan and Lucassen, chapter 10) • Genetic and genomic testing in healthcare: practical and ethical aspects (Strachan and Lucassen, chapter 11)