



Syllabus Master's Degree Course in Medicine and Surgery

THE CELL: MOLECULES AND PROCESSES

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

Teachers

Subject	Academic credits (CFU)	Lecturer
Molecular biology II	1	IRACI Nunzio
Applied biology II	4	RAGUSA Marco
Medical genetics II	2	ROMANO Corrado

Learning outcomes

Subject	Learning outcomes
Molecular biology II	<p>By the end of the course, students are expected to:</p> <ul style="list-style-type: none">• 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 courses of Molecular Biology, Applied Biology and Medical Genetics.
Applied biology II	<p>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:</p> <ul style="list-style-type: none">• knowledge of cell functions• 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, especially the role of non-coding RNAs• understanding of the principles of cellular and molecular bases of cancers
Medical genetics II	<p>By the end of the course, students are expected to:</p> <ul style="list-style-type: none">• Demonstrate an understanding of the pathogenic role of genetic variation, including its impact on disease development and progression.• Identify and assess disease genes and genetic susceptibility factors related to complex diseases, utilizing various research and diagnostic techniques.

	<ul style="list-style-type: none"> • Apply genetic approaches to the development and treatment of diseases, including the use of genetic therapies and targeted interventions. • Analyze and discuss the principles and key concepts of cancer genetics and genomics, including the genetic basis of various types of cancer. • Evaluate and implement genetic and genomic counseling and testing strategies within a healthcare context, considering ethical and clinical implications for patients and families.
--	---

Prerequisites

Subject	Prerequisites
Molecular biology II	Basic notions of Biochemistry and Molecular Biology.
Applied biology II	Basic notions of Biology and Genetics.
Medical genetics II	Attainment of the educational objectives set by prerequisite courses.

Course contents

Subject	Course contents
Molecular biology II	<ul style="list-style-type: none"> • Manipulation of nucleic acids: electrophoresis and nucleic acid hybridization • Recombinant DNA: molecular cloning • PCR, RT-PCT and qPCR • DNA sequencing and NGS • Gene expression and protein-nucleic acid interactions: chromatography; western blot; EMSA; ELISA; immunoprecipitation; chromatin immunoprecipitation • Transgenic organisms
Applied biology II	<p>Cellular Movement: Motility and Contractility</p> <ul style="list-style-type: none"> • Microtubule-based movement inside the cells: kinesins and dyneins • Microtubule-based cell motility: Cilia and flagella • Microfilament-based movement inside the cells: Myosin • Microfilament-based motility: muscle cells • Microfilament-based motility in nonmuscle cells <p>Extracellular Structures, Cell Adhesion, and Cell Junctions</p> <ul style="list-style-type: none"> • Cell – cell junctions • The extracellular matrix of the animal cells <p>Epigenetics</p> <ul style="list-style-type: none"> • Chromatin modification • DNA methylation • Non-coding RNAs and gene expression regulation <p>Signal Transduction Mechanisms</p> <ul style="list-style-type: none"> • Chemical signals and cellular receptors • G-protein coupled receptors • Enzyme-coupled receptors • Synaptic transmission • Hormones and other long-range signals

	<p>The Cell Cycle and Mitosis</p> <ul style="list-style-type: none"> • Nuclear and cell division • Regulation of cell cycle • Growth factors and cell proliferation • Sexual Reproduction and Meiosis <p>Apoptosis</p> <ul style="list-style-type: none"> • Death signals and survival factors • Apoptotic pathways • Stem cells <p>Cancer cells</p> <ul style="list-style-type: none"> • How cancers arise • How cancers spread • What causes cancers • Oncogenes and Tumor suppressor genes
<p>Medical genetics II</p>	<p>The pathogenic role of genetic variation</p> <ul style="list-style-type: none"> • an overview of how genetic variation results in disease • pathogenic nucleotide substitutions and tiny insertions and deletions • pathogenic variation in short tandem repeat copy number • pathogenic variation in long tandem repeats and interspersed repeats • chromosome abnormalities and copy number variants • molecular pathology of mitochondrial disorders • effects on phenotype of pathogenic variation in nuclear DNA • the protein structure perspective of molecular pathology <p>The identification of disease genes and genetic susceptibility to complex diseases</p> <ul style="list-style-type: none"> • the identification of genes in monogenic disorders • the identification of genetic susceptibility to complex diseases • the genetic architecture of complex disease and the contribution of environment and epigenetics <p>Genetic approaches to the treatment of diseases</p> <ul style="list-style-type: none"> • 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 <p>Cancer genetics and genomics</p> <ul style="list-style-type: none"> • fundamental characteristics and evolution of cancer • oncogenes and tumour suppressor genes • genomic instability and epigenetic dysregulation in cancer • genome-wide studies of cancers • genetic inroads into cancer therapy <p>Genetic and genomic testing in healthcare: practical and ethical aspects</p> <ul style="list-style-type: none"> • overview of genetic testing • genetic testing for chromosome abnormalities and pathogenic structural variation • genetic testing for pathogenic point mutations and DNA methylation • genetic counselling and testing services: practical applications

	<ul style="list-style-type: none"> ethical, legal, and societal issues (ELSI) in genetic testing and counselling
--	---

Assessment methods

Subject	Assessment methods
Molecular biology II	<p>Since it is an annual course, the exam will be held in the second semester along with the first 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.</p> <p>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 of the course modules; ii) the clarity of presentation; iii) the property of medical-scientific language. The assessment of learning can also be conducted remotely if the conditions necessitate it.</p> <p>For the assignment of the final grade, the following parameters will be considered:</p> <ul style="list-style-type: none"> 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. <p>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.</p>
Applied biology II	
Medical genetics II	

Examples of common questions and/or exercises

Subject	Examples of common questions and/or exercises
Molecular biology II	<ul style="list-style-type: none"> The mechanism of PCR Restriction endonucleases and the molecular cloning Strategy of DNA sequencing
Applied biology II	<ul style="list-style-type: none"> Biological meaning of apoptosis The role of non-coding RNAs in post-transcriptional regulation

	<ul style="list-style-type: none"> • The role of G-protein coupled receptors • Control of cell cycle • Molecular bases of cancer development
Medical genetics II	<ul style="list-style-type: none"> • The pathogenic role of genetic variation • The identification of disease-causing genes • The impact of environment and epigenetics in the phenotype • Cancer genetics • Genetic services • Genetic treatment of the diseases

Reference texts

Subject	Textbooks
Molecular biology II	<ul style="list-style-type: none"> • 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. <p>Any additional educational material (slides, videos, handouts, etc.) will be distributed or indicated during the lessons.</p>
Applied biology II	<ul style="list-style-type: none"> • Becker's World of the Cell, Global Edition, Hardin and Lodolce, tenth edition. Pearson. <p>Any additional educational material (slides, videos, handouts, etc.) will be distributed or indicated during the lessons.</p>
Medical genetics II	<ul style="list-style-type: none"> • 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. <p>Any additional educational material (slides, videos, handouts, etc.) will be distributed or indicated during the lessons.</p>

Course format

Subject	Textbooks
Molecular biology II	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 II	
Medical genetics II	

Attendance

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

Course schedule

Subject	Textbooks
Molecular biology II	<ul style="list-style-type: none"> 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 II	<ul style="list-style-type: none"> Cellular Movement: Motility and Contractility: Becker's World of the Cell (chapter 14) Extracellular Structures, Cell Adhesion, and Cell Junctions: Becker's World of the Cell (chapter 15) 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 Cancer cells: Becker's World of the Cell (chapter 26)
Medical genetics II	<ul style="list-style-type: none"> 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)