Degree Course in Dentistry and Dental Prosthetics

Course: Biology and Genetics
CFU Number: 11
Coordinator of the integrated course: Cinzia Ciccacci; **email:** cinzia.ciccacci@unicamillus.org

Module: Applied Biology
SSD Course: BIO/13
CFU Number: 8
Professor names: Bruni Emanuele (6 CFU), Paolo Chieffi (2 CFU)
Professor email: paolo.chieffi@unicamillus.org, emanuele.bruni@unicamillus.org

Module: Medical Genetics
SSD Course: MED/03
CFU Number: 3
Professor names: Cinzia Ciccacci
Professor email: cinzia.ciccacci@unicamillus.org

PREREQUISITES

There are no prerequisites, but it would be desirable that student already knows basic biology elements, such as the gene structure, DNA replication, concepts of meiosis and mitosis.

OBJECTIVES

The integrated course of Biology and Genetics will provide the student with the basis for understanding the biological processes and the diseases causing mechanisms. The Biology teaching will provide knowledge useful to distinguish living systems, with particular reference to: the structural and functional characteristics of the cell; the study of the processes and the most important cellular activities; the study of the molecular mechanisms underlying the regulation of gene expression in relation to the cell differentiation process, as a differential expression of the genetic heritage.
The Medical Genetics teaching will provide to students the knowledge on the main notions on inheritance of monogenic, chromosomal and multifactorial diseases. At the end of the course the student will be able to distinguish the main classes of genetic diseases and to recognize the modes of transmission of hereditary diseases.

LEARNING OUTCOMES

At the end of the course, the student must have acquired:
Knowledge and understanding
- The knowledge of the principles of classification of living beings.

- The knowledge of the main characteristics and differences between prokaryotic and eukaryotic cells.
- The knowledge of the main cellular compartments and their function.
- The knowledge of the general principles of cellular metabolism.
- The knowledge of the molecular basis of the transmission of hereditary characters.
- The knowledge of the molecular basis of gene expression.
- The knowledge of the mechanisms underlying cell differentiation.
- The understanding of the molecular and cellular mechanisms underlying diseases
- The knowledge of the main inheritance models of monogenic, chromosomal and multifactorial diseases
- The understanding of how to reconstruct family pedigrees and to calculate disease recurrence
- The understanding of the major kinds of genetic testing and their proper use.

Applying knowledge and understanding
- The ability to describe biological processes and the mechanisms that regulate them
- The understanding of the molecular and cellular mechanisms underlying diseases
- The capacity to analyse family pedigrees and clinical and molecular genetic data useful for genetic counselling
- The ability to calculate disease recurrence risk

Communication skills
- The ability to adequately describe a biological phenomenon by demonstrating an appropriate scientific language
- The use of correct genetic terminology
- The ability to describe the main models of inheritance and the recurrence risk

Making judgements
- The ability to synthesize and correlate the various topics
- A critical ability on the use of genetic tests for the molecular diagnosis of monogenic and chromosomal diseases or for the evaluation of genetic susceptibility to complex diseases

COURSE SYLLABUS: Biology

- Main characteristics of living systems: fundamental properties of cells; the cell theory; organization, structure, complexity and main differences between prokaryotic and eukaryotic cells; structure and functions of cellular organelles; definition and classification of viruses and stages of viral infection.

- Chemical basis of life: main chemical bonds characterizing living matter; structure and function of the four biological molecules (carbohydrates, lipids, nucleic acids and proteins); definition of enzymes as biological catalysts.

- Plasma membrane: properties and functions, modes of transport of small molecules across the
plasma membrane (simple diffusion, facilitated diffusion, active transport).

- **Internal organization of the cell:** intracellular compartments and protein sorting, intracellular
membrane traffic, organelles. The cytoskeleton and cell motility. Nuclear compartment, structure
and functions. The different levels of chromatin condensation.
- **Energy conversion:** Glycolysis and fermentation (outline). Mitochondria and Chloroplasts, structure and function.
- **Molecular basis of hereditary information**: DNA replication. DNA repair and its correlation with
human diseases.
- **RNA, structure and function:** Main types of cellular RNAs and differences with respect to DNA in
terms of molecular size, stability and biological functions. Transcription and RNA maturation.
- **Genetic Code and its properties. Protein synthesis:** How cells read the genome. Main posttranslational
modifications of the polypeptide chains.
- **Post-synthetic fate of proteins, endomembranes and membrane traffic:** Endocytosis, pinocytosis,
phagocytosis and receptor-mediated endocytosis (LDL).
- **Control of gene expression*:*** Molecular mechanisms that create specialized cell types. Functional
organization of the eukaryotic genome. Histone code. Transcriptional control, role of chromatin
condensation and of the degree of DNA methylation.
- **Apoptosis and Cancer:** Tumor suppressors and proto-oncogenes.
- **Cell Signaling:** Exchange of chemical signals through receptor proteins. The key role played by
protein kinases in this process.
- **The interactions between cells and their environment**: Adhesion molecules and extracellular
matrix.
- **Cell Cycle:** The cell cycle control system.
- **Mitosis and Meiosis:** principles of chromosome dynamics during mitosis and meiosis; molecular
mechanisms of genetic recombination.
- **Gene mutations:** mutations by substitution, insertion or deletion of nucleotides. Spontaneous and
induced mutations. Chemical and physical mutagens. DNA repair systems for single or double
stranded DNA damage.
- **Mobile genetic elements and evolution of the genomes**

<u>COURSE SYLLABUS:</u>Medical Genetics

- Basic Genetics: Definitions of Key Terms: gene, locus, allele, genotype, phenotype, haplotype, homozygous, heterozygous, haploid, diploid, dominance, recessivity, codominance, mutation, polymorphism.
- Principles of Genetic Transmission: Mendel's Genetic Hypothesis, The Monohybrid and Dihybrid Crosses, Segregation in Human Pedigrees, Blood groups Genetics
- Monogenic Inheritance Models: Autosomal inheritance,Autosomal recessive inheritance, X-linked inheritance
- Genetic Risk calculation and pedigrees analyses. Hardy-Weinberg equilibrium.
- Chromosomes: Structure and Analysis, Chromosomes Pathologies
- Genomic Imprinting
- X-chromosome inactivation
- Mitochondrial inheritance: mitochondrial DNA, pattern of inheritance
- Multifactorial Inheritance and Common Diseases: polimorphisms, susceptibility genes, gene-environment interaction, association studies
- Pharmacogenomics and Personalised Medicine
- Genetic tests and Counselling.
- Prenatal and postnatal testing
- Examples of genetic diseases: BRCA1 and breast cancer, Cystic fibrosis, sex development disorders, dinamic mutations disorders, genes and dental disorders
- Epigenetics and nutrigenetics. Outlines

COURSE STRUCTURE

The course is divided in frontal lectures, 80 hours in Biology and 30 hours in Medical Genetics. The teachers use educational tools such as presentations organized in powerpoint files with explanatory diagrams, illustrations and images to describe the various topics of the course. Moreover, Medical Genetics Lectures will include also interactive lessons with exercises. The attendance at lectures is mandatory.

COURSE GRADE DETERMINATION

Student learning will be assessed through a written and oral exam.
The Biology written test is mandatory to be admitted to the oral exam, but it does not contribute to the
final score. The written test is organised into 30 questions with multiple-choice answers in which only one is correct. Each correct answer is given a score of 1. There is no penalty for wrong or not given answers. To access the oral test, the student must have obtained at least 18/30. Notably, the written test is a selection test; in the oral test the student is given the opportunity to demonstrate his preparation by discussing the topics of the course, to reason on issues related to biology demonstrating that he has acquired the ability to express himself in a suitable scientific language. The final evaluation will be based mainly on the outcome of the oral test.

The Medical Genetics exam consists in a mandatory written test, followed by an optional oral. The written test will consist of 30 questions with multiple choice answers, for each correct answer 1 point will be assigned. The final score of the written test will be given by the sum of

the partial scores assigned to correct answers. There is no penalty for wrong or not given answers. To access the oral exam student must have at least a minimum of 18 points at the written test (18/30). The oral exam is optional. The minimum score to pass the exam is 18/30.

READING MATERIALS/BOOK LIST:

BIOLOGY: Karp's Cell and Molecular Biology: Concepts and Experiments, Gerald Karp; Janet Iwasa; Wallace Marshall

Medical Genetics: Lectures in pdf format will be provided to students. Recommended books: “Medical Genetics” by Lynn Jorde, John Carey, Michael Bamshad. Edited by Elsevier