

**COURSE OF MEDICINE AND SURGERY**  
**Student Handbook a.y. 2012-2013**

**BIOLOGY AND GENETICS**

I Year	Scientific Field	DISCIPLINE	TUTOR
<b>Biology and Genetics</b>  ECM 10  Coordinator <b>Maria Giulia Farace</b>	BIO/13	<i>Applied Biology</i>	<b>Farace Maria Giulia</b>
	BIO/13	<i>Applied Biology</i>	<b>Achsel Tilmann</b> <i>Catholic Univ. of Leuven</i>
	BIO/13	<i>Applied Biology &amp; Molecular Genetics</i>	<b>Bagni Claudia</b>
	BIO/13	<i>Genetics</i>	<b>Ciafrè Silvia Anna</b>
	MED/03	<i>Medical Genetics</i>	<b>Giardina Emiliano</b>

**Specific aims** The aim of the integrated course of Biology and Genetics is to learn the experimental method and its application to the study, at level of structure and function of:

- the common mechanisms regulating cell function and activity and the way in which cells interacts with each other;
- the molecular mechanisms responsible for the diversification of biological units, in relation to their internal structure and compartmentalization, differential gene expression both at level of single organism (differentiation) and during evolution;
- the fundamental molecular mechanisms of heredity in living cells and populations.

**PROGRAM**  
**Cell Biology**

- Characteristics of the living cells. Cellular theory. Principles of classification of living organisms.
- Chemistry of living matter. Macromolecules: structure, shape and information.
- Molecular interactions in biological entities. Viruses: definition as intracellular parasites, classification on the basis of the type of genomic nucleic acid and the type of infected cell. Lytic and lysogenic cycles.
- Prokaryotic and eukaryotic cell models: classification and major structural differences.
- Plasma membranes (properties and functions) and cell walls: physical and chemical properties of the membranes in relation to their lipid composition; intrinsic and extrinsic membrane proteins and topological organization of proteins in the lipid bilayer; main functions of membrane proteins; modes of transport of small molecules across the plasma membrane (simple diffusion, facilitated diffusion, active transport).
- Glycolysis and fermentation (outline). Mitochondria and cellular respiration. Chloroplasts and photosynthesis (outline). Correlation between energy conversion and cellular structures. Characteristics of mitochondrial membranes, mitochondria and the evolution of the eukaryotic cell. Mutual dependence between photosynthesis and cellular respiration.
- Nuclear compartment (carioteca, nucleolus, chromatin): structure and functions. The different levels of chromatin condensation.
- Molecular basis of the hereditary information: correlation between structure and function of nucleic acids.
- DNA replication. Telomerase. DNA repair and its correlation with human diseases. The most frequent types of errors that can occur under physiological conditions during DNA metabolism, and the main mechanisms of DNA repair in eukaryotic cells.
- RNA, structure and function: main types of cellular RNAs and differences with respect to DNA in terms of molecular size, stability and biological functions.



## BIOLOGY AND GENETICS

- Transcription and RNA maturation.
- Genetic code and its properties. Protein synthesis.
- Cellular compartments: biogenesis and maintenance. Post-synthetic fate of proteins, endomembranes and membrane flow. Exocytosis and endocytosis. Main post-translational modifications of the polypeptide chains. Fission and fusion of membranes. Peroxisome biogenesis. Mechanisms of various forms of endocytosis: pinocytosis, phagocytosis and receptor-mediated endocytosis (LDL).
- Development and cell differentiation: cell differentiation as the differential expression of a single genome common to all cells of the same organism.
- Functional organization of the eukaryotic genome. Histone code. Regulatory sequences, DNA / protein interactions.
- Molecular mechanisms of the regulation of gene expression. Transcriptional control: role of chromatin condensation and of the degree of DNA methylation. In cis elements and in trans factors. Translational and post-transcriptional control.
- Cell cycle and its control. Apoptosis. Tumor suppressors and protooncogenes.
- Cellular communication and signal transduction. 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(Histology)
- Cytoskeleton and cell motility(Histology).

### PROGRAM Genetics

- Mitosis and Meiosis: principles of chromosome dynamics during mitosis and meiosis; similarities and differences between the two processes. Molecular mechanisms of genetic recombination. Concepts of homologous chromosomes, aploidy and diploidy.
- The chromosomes. Methods for chromosome analysis. Normal karyotype and chromosomal heteromorphisms.
- Mendelian inheritance. Mendel's experiments and the concept of segregation of characters. Basic concepts of probability. Alleles and loci, homozygosity and heterozygosity, dominance and recessivity, incomplete dominance, codominance. Pleiotropy. Multiple alleles. Essential genes and lethal alleles.
- 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.
- Mitochondrial inheritance: relevance for human phylogenetic tree reconstruction.
- Population genetics: Hardy-Weinberg equilibrium and theoretical implications for understanding the mechanisms of biological evolution. Calculation of gene and genotype frequencies for two-allele systems in genetic counseling.
- Mobile genetic elements and evolution of the genome.

### PROGRAM Medical Genetics

- Autosomal dominant and recessive inheritance
- Sex-linked and non traditional modes of inheritance
- Clinical cytogenetics: the chromosomal basis oh human diseases
- Multifactorial inheritance and common diseases
- Genetic testing and genetic counseling

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**TEST Books**

**Cell biology :**

“ Molecular Biology of the Cell”, Bruce Alberts et al., Garland Science, 2007

**Genetics andMolecular genetics:**

“ iGenetics: a molecular approach, Peter J. Russell, Benjamin/Cummings Pub. Co

**Medical Genetics:**

“Medical Genetics” Jorde, Carey, Bamshad. Ed. MOSBY Elsevier

**METHOD of TEST**

Oral exam.

**TEST COMMISSION**

<b>Maria Giulia Farace</b> ( <i>President</i> )	
<b>Bagni Claudia</b>	
<b>Silvia Anna Ciafrè</b>	
<b>Emiliano Giardina</b>	
<b>Tilmann Achsel</b> Catholic University of Leuven	



**Tutor**

<b>Farace Maria Giulia</b> ( <i>Coordinator</i> )	<a href="mailto:mariagiulia.farace@uniroma2.it">mariagiulia.farace@uniroma2.it</a>	06 7259 6087
<b>Bagni Claudia</b>	<a href="mailto:Bagni.Claudia@cme.vib-kuleuven.be">Bagni.Claudia@cme.vib-kuleuven.be</a>	06 7259 6063
<b>Ciafrè Silvia Anna</b>	<a href="mailto:ciafre@uniroma2.it">ciafre@uniroma2.it</a>	06 7259 6059
<b>Giardina Emiliano</b>	<a href="mailto:emiliano.giardina@uniroma2.it">emiliano.giardina@uniroma2.it</a>	06 7259 6072
<b>Achsel Tilmann</b> Catholic University of Leuven	<a href="mailto:Tilmann.Achsel@cme.vib-kuleuven.be">Tilmann.Achsel@cme.vib-kuleuven.be</a>	06 7259 6063