I year (2nd semester)	Scientific Field	BIOLOGY AND GENETICS	TUTOR	ECTS
	BIO/13	Applied Biology	Ciafrè Silvia Anna	7
	BIO/13	Applied Biology	Farace Maria Giulia	2
CIAFRE' S.A. **COORDINATOR**	MED/03	Medical Genetics	Botta Annalisa	1
			TOT	10

PROGRAM **CELL BIOLOGY**	- CHARACTERISTICS OF LIVING CELLS. Cellular theory. Classification principles 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.
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 proto-oncogenes.
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, haploid and diploid. - 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 of human diseases. - Multifactorial inheritance and common diseases. - Genetic testing and genetic counseling.

TEXTBOOKS	Cell biology: “ Molecular Biology of the Cell”, VI edition, Bruce Alberts et al., Garland Science, 2014. Genetics and Molecular genetics: “Principles of Genetics” VI edition, Snustad and Simmons, Wiley ed. Medical Genetics: “Medical Genetics” Jorde, Carey, Bamshad. Ed. MOSBY Elsevier
EXAM METHOD	Oral exam.

EXAM COMMISSION	The Coordinator, full Professors of the disciplines, Professors of similar disciplines, Specialists of the subject, compose the exam Commission of the Integrated Course.

Ciafrè Silvia Anna, President
Farace Maria Giulia
Botta Annalisa

CONTACTS

Ciafrè Silvia Anna	ciafre@uniroma2.it	0672596059
Botta Annalisa	botta@med.uniroma2.it	0672596078
Farace Maria Giulia	mariagiulia.farace@uniroma2.it	

PREREQUISITES: Previous knowledge and competence in the following subjects: Chemistry and Introductory Biochemistry, Physics and Statistics.

The specific learning outcomes of the program are coherent with the general provisions of the Bologna Process and the specific provisions of EC Directive 2005/36/EC. They lie within the European Qualifications Framework (Dublin Descriptors) as follows:

1. **Knowledge and Understanding**

- Demonstrate a comprehensive theoretical knowledge of the main biological and genetic processes and provide a foundation in the basic principles.
- Explain the relationships between form and function of biological structures at the molecular, cellular, organismal, population, and ecosystem levels of the biological hierarchy.
- Identify the structural components of the cell and define the main processes of cell growth, metabolism, communication, regulation, replication and death.
- Describe the concept of chromosome dynamics and the main mechanisms of inheritance. Recognize the consequences related to an alteration of function, applying them to the clinical setting.
- Explain the importance of biodiversity at the genetic, organismal, community, and global scales.

2. **Applying Knowledge and Understanding**

- Identify and recognize the proper diagnostic techniques to utilize for any particular topic of examination; giving a comprehensive description of all the available possibilities.
- Recognize the importance of genetic testing and genetic counseling in the general population. Practice on main genetic problems and learn how to construct a pedigree.
- Learn to interpret scientific data and conditions and acquire problem-solving abilities, which combine a scientific and ethical approach.
- Learn the practical aspects related to investigative tests and how to perform them.

3. **Making Judgements**

- Recognize the importance of an in-depth knowledge of the topics consistent with a proper medical education.
- Identify the benefits and adverse effects of any diagnostic and therapeutic interventions.
- Identify the fundamental role of a proper theoretical knowledge of the subject in the clinical practice.

4. **Communication Skills**

- Present the topics orally in an organized and consistent manner.
- Use of proper scientific language coherent with the topic of discussion.

5. Learning Skills

- Identify the possible use of the acknowledged skills in the future career.
- Assess the importance of the acquired knowledge in the overall medical education process.