BIOMEDICAL SCIENCES II (ML000025)

1. lingua insegnamento/language

Inglese.

2. contenuti/course contents

Coordinatore/Coordinator: Prof. ARCOVITO ALESSANDRO

Anno di corso/Year Course: 2021/2022

Semestre/Semester: Secondo

CFU/UFC: 13

Moduli e docenti incaricati /Modules and lecturers:
- BIOCHEMISTRY (ML000028) - 6.66 cfu - ssd BIO/10
Prof. Federica Iavarone, Alessandro Arcovito
- BIOCHEMISTRY PRACTICALS (ML000029) - 0.34 cfu - ssd BIO/10
Prof. Francesca Sciandra, Manuela Bozzi, Manuela Bozzi, Giuseppina Nocca
- CELLULAR BIOLOGY II (ML000026) - 1.5 cfu - ssd BIO/13
Prof. Wanda Lattanzi
- CELLULAR BIOLOGY II PRACTICALS (ML000027) - 0.5 cfu - ssd BIO/13
Prof. Lorena Di Pietro, Valentina Saccone, Daniela Palacios Garcia
- HUMAN GENETICS (ML0377) - 3 cfu - ssd MED/03
Prof. Pietro Chiurazzi, Giuseppe Marangi
- MOLECULAR BIOLOGY (ML0367) - 1 cfu - ssd BIO/11
Prof. Andrea Urbani

3. testi di riferimento/bibliography

CELLULAR BIOLOGY II

Karp G, “Cell and Molecular Biology – Concepts and Experiments” 8th edition, Wiley, 2016 (latest available edition)

For additional consultation: Alberts B, et al. Essential Cell Biology, 4th Edition, Garland Science - Taylor & Francis Group, 2013.

BIOCHEMISTRY and MOLECULAR BIOLOGY

David L. Nelson, Michael M. Cox, Lehninger Principles of Biochemistry, VIII edition, McMillan Learning

HUMAN GENETICS

Emery's Elements of Medical Genetics and Genomics. 16th Edition (Turnpenny, Ellard, Cleaver) – 16th edition, Elsevier 2020 (ISBN: 9780702079665). Chapters: 1,2,3,4,5,6,7,8,10,12.

Additional material will be provided (i.e. scientific articles and appropriate telematic sources) to complete, elaborate and update the contents discussed in each module.

4. obiettivi formativi/learning objectives

The whole course is aimed at giving the student a deep knowledge of the main metabolic and molecular processes that occur in the human organism along with the basics of heredity and the principles of human and medical genetics. Moreover, insights into the modern molecularbiology techniques and their relevance for modern medicine will be provided.

5. prerequisiti/PREREQUISITES

In order to better understand the topics of the course and to formally register for the exam session, students must have attended and passed the exam of Basic Sciences.

6. metodi didattici/teaching methods

At the end of the integrated course the student must demonstrate that he/she has reached the following objectives:

Knowledge and understanding abilities – demonstrate the know-how of the different metabolic and molecular processes occurring in the eukaryotic cells enabling intercellular communications and homeostasis, along with their implication in genetic disorders.

Applied knowledge and understanding skills – demonstrate to adequately interpret the importance of molecular mechanisms and hereditary patterns that may be altered in human diseases.

Personal judgement – the student must properly integrate the knowledge and skills learned to develop autonomous abilities to identify the fundamental pathways associated with physiological mechanisms, relevant for the application in the medical field.

Communication skills – be able to communicate scientific and applicative content in a clear and unambiguously way, using an appropriate technical language and explaining their personal conclusions, as well as the knowledge and rationale underlying them, to specialists and non-specialist interlocutors.

Learning ability – be able to keep up-to-date and expand their knowledge by autonomously drawing on scientific texts and articles, and scientific databases (NCBI/PubMed, MedLine,).

The course is organized into lectures and practical sessions covering the topics included in all the teaching modules to provide the basic elements of Cell Biology, Biochemistry, Human Genetics and Molecular Biology. The teaching methods implement active learning activities, such as problem-based learning, self-learning, case studies and experimental activities.

The adopted teaching methods allow students to pursue the learning objectives:

Knowledge and understanding abilities – all the topics listed in the program will be treated during frontal lectures and carefully explained with tutorials and exhaustive explanations, in order to provide students the appropriate knowledge and learning method.

Applied knowledge and understanding skills – the interactive teaching methods, including “case study”, practical examples and laboratory practice exercises, will allow students to learn the applicative potential of the treated topics.
Personal judgement – the active learning modalities will enable students to independently formulate ideas and use critical reasoning.

Communication skills – the active learning modalities will stimulate active and constant interaction with the instructors, towards the progressive development of focused communication skills for the presentation of treated topics through a correct scientific language.

Learning ability – additional material (i.e. scientific articles and appropriate telematic sources) will support students in their individual and autonomous study.

7. altre informazioni/other informations

The instructors are available for further clarifications and discussions regarding the learning modules’ topics and any possible related issue, also beside the classes timetable by appointment.

8. modalità di verifica dell’apprendimento/ methods for verifying learning and for evaluation

The exam is composed of a written test comprising 50 multiple-choice quizzes divided into 2 blocks:

1. Biochemistry (26 quizzes)

2. Molecular Biology (4 quizzes), Cellular Biology II (8 quizzes), Human Genetics (12 quizzes), for a total of 24 quizzes

For each quiz a single correct answer is possible. Each correct answer correspond to a score= 1, wrong/no answer correspond to score= 0. To pass the written test the student needs to answer correctly to at least 15 quizzes in module 1 and 15 quizzes in module 2.

The final score obtained in the written test is calculated according to the following scale:

30/50=18
31/50=18
32/50=19
33/50=19
34/50=20
35/50=21
36/50=21
37/50=22
38/50=22
39/50=23
40/50=24
41/50=25
42/50=25
43/50=26
44/50=27
45/50=27
46/50=28
47/50=28
48/50=29
49/50=30
50/50 =30 with honors

The minimum score that needs to be achieved to pass the written test is 18. Passing the written test will give access to a brief oral examination. During the oral test the students will be asked an overall number of 4 questions, each scoring in the -1/+1 range. The final score achieved in the oral part will be then -4/+4 and will be added to the score achieved in the written test to calculate the final score. The student will be able to obtain the maximum final score (30/30 with honors) only if he/she achieved a minimum score of 27/30.

The objective of the evaluation system is to verify:

the knowledge and understanding of the different aspects that are discussed in each module during the course (knowledge and understanding);the ability to connect theoretical concepts and practical problems regarding the different aspects discussed during the course (Applied knowledge and understanding);

- the ability to integrate the themes treated in the different modules of the course (Personal judgement);

- the development of communication skills for the presentation of treated topics through a correct scientific language (Communication skills);

- the ability to autonomously delve into specific biomedical topics (Learning ability).

9. programma esteso/program

CELLULAR BIOLOGY II

DNA and the genetic code. Eukaryotic genes and genome organization. DNA replication in prokaryotes and eukaryotes. DNA repair mechanisms. The RNA world. RNA transcription, post-transcriptional modifications. Noncoding RNAs. Gene expression regulation. Protein translation, post-translational regulation of protein expression and function. Signal transduction and intracellular signalling: introduction to main signalling cascades and biological pathways.

CELLULAR BIOLOGY II PRACTICALS:

The Cellular Biology II Practical sessions are based on laboratory practice exercises to study gene expression in *in vitro* cell culture models, applying the following techniques:

1. cDNA synthesis: reverse transcription reaction;
2. Polymerase Chain Reaction (PCR);
3. Real-Time PCR.

BIOCHEMISTRY

Enzymes. Enzymatic catalysis and regulation. Passive and active transport across membranes. Introduction to metabolisms. Glycolysis and its regulation. The pentose phosphate pathway and the additional pathways in carbohydrate metabolism. Glycogen synthesis and breakdown. Gluconeogenesis and other carbohydrate biosynthetic pathways. The citric acid cycle and its regulation. Mitochondrial ATP synthesis and electron transport. Oxidative Phosphorylation. Metabolism of nucleic acid, anabolism and catabolism of purines and pyrimidines. Synthesis and degradation of lipids. Regulation of fatty acid metabolism. Synthesis of other lipids. Cholesterol metabolism. Synthesis and degradation of amino acids. Urea cycle. Nitrogen metabolism, anabolism and catabolism of glutathione, creatine, active amine, NO and heme groups. Hormones

and signal transduction. Bioenergetics and regulation of fuel metabolism.

BIOCHEMISTRY PRACTICALS:

Determination of the protein concentration in a cellular extract using the Bradford Method. Determination of the enzymatic activity in vitro.

MOLECULAR BIOLOGY

Nucleotides and Nucleic Acids structures: Nucleic Acid Structure, Nucleic Acid Chemistry, Other Functions of Nucleotides; DNA- Based Information Technologies: DNA Cloning: The Basics, From Genes to Genomes, From Genomes to Proteomes, Proteomics investigations; Structure to function analysis: from gene to protein structures, bioinformatics analysis of gene and protein sequences, developing and screening expression libraries, recombinant protein expression and purification.

HUMAN GENETICS

Structure and functions of genes and human genome: nucleic acids, basic structure of gene, organization of the human genome, central dogma of molecular biology, mRNA splicing, alternative splicing, genetic code. Genetic variants: large-scale and small-scale variants; substitutions, deletions, duplications, insertions, microsatellites, inversions; inherited variants and “de novo” mutations; germinal and somatic mosaicism; missense, nonsense, frameshift and in-frame variants; silent, loss-of-function, and gain-of-function variants; dominant negative, haploinsufficiency; frequency of genetic variants. Mendel’s laws of inheritance: recessive and dominant inheritance, allele segregation, independent assortment. Genealogical data and family trees. Mendelian inheritance of monogenic disorders: autosomal dominant/recessive, X-linked. Incomplete penetrance and variable expressivity. Exceptions to Mendel’s laws: codominance, random monoallelic expression (RME) and parental-specific monoallelic expression (imprinting), X-inactivation, linkage and crossing-over. Cytogenetics: human chromosomes and karyotype; mitosis, meiosis and crossing over; chromosome number anomalies, polyploidy, aneuploidy, trisomies and monosomies; chromosome structure anomalies (aneusomies), balanced and unbalanced; deletions, duplications and amplifications, reciprocal and Robertsonian translocations, insertions, isochromosomes, ring chromosomes, copy number variants; chromosome analysis, array-CGH, FISH analysis. Genetic markers and Linkage studies. Techniques of DNA and RNA analysis, Next-Generation Sequencing, Sanger sequencing, MLPA. Genetic Databases and homology searches.