GENE REGULATION AND CELL METABOLISM (MZ000005)

1. LANGUAGE

English.

2. COURSE CONTENTS

Coordinator: Prof. FRANCESCO CECCONI
Year Course: I
Semester: 2nd semester
CFU/UFC: 11
Modules and lecturers:
- BIOCHEMISTRY (MZ000061) – 5,50 CFU - SSD BIO/10
  Prof. Matteo Bordi, Prof. Viviana Greco
- BIOCHEMISTRY PRACTICALS (MZ000062) – 0,50 CFU - SSD BIO/10
  Prof. Emanuele Panatta
- CELLULAR BIOLOGY 2 (MZ000063) - 2 CFU - SSD BIO/13
  Prof. Andrea Papait
- CELLULAR BIOLOGY PRACTICALS (MZ000064) – 0,50 CFU - SSD BIO/13
  Prof. Andrea Papait
- HUMAN GENETICS (MZ000066) – 1,50 CFU - SSD MED/03
  Prof. Eugenio Sangiorgi
- MOLECULAR BIOLOGY (MZ000065) - 1 CFU - SSD BIO/11
  Prof. Francesco Cecconi

3. BIBLIOGRAPHY

CELLULAR BIOLOGY 2

Alberts B, et al. Molecular Biology of the Cell, 7th Edition, Garland Science - Taylor & Francis Group, 2022.

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

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. 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 molecular biology techniques and their relevance for modern medicine will be provided.

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

Knowledge and understanding abilities (Dublin 1) – 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 (Dublin 2) – demonstrate to adequately interpret the importance of molecular mechanisms and hereditary patterns that may be altered in human diseases.

Personal judgement (Dublin 3) – 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 (Dublin 4) – 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 (Dublin 5) – 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).

5. PREREQUISITES

Students **must have successfully completed and attended the Basic Sciences** examination in order to formally register for the exam session and gain a more comprehensive understanding of the course subject.

6. TEACHING METHODS

The course is structured with lectures and practical activities that encompass the subjects covered in all teaching modules, aiming to deliver fundamental concepts in Cell Biology, Biochemistry, Human Genetics, and Molecular Biology. The teaching approaches incorporate active learning strategies, including problem-based learning, self-directed learning, case studies, experimental activities, and they may include forms of interactive lecturing.

Knowledge and understanding abilities (Dublin 1) – All the topics outlined in the program will be covered through face-to-face lectures and supplemented with tutorials, comprehensive explanations. and forms of interactive online lecturing. This approach aims to provide students with the necessary knowledge and effective learning strategies.

Applied knowledge and understanding skills (Dublin 2) – The use of interactive teaching methods, such as “case study”, practical examples, and laboratory practice exercises, will facilitate students' understanding of how the topics can be applied in real-life situations.

Personal judgement (Dublin 3) – The active learning modalities will empower students to

independently generate ideas and employ critical reasoning skills.

Communication skills (Dublin 4) – The active learning methods will encourage continuous and engaged engagement with teachers, leading to the gradual improvement of focused communication skills for presenting issues using appropriate scientific language.

Learning ability (Dublin 5) – Additional material such as scientific papers and relevant online sites, will assist students in their independent and self-directed study.

7. 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 previously arranged by e-mail communication.
Please DO NOT refer to instructors for seeking technical assistance on class attendance or exam registration as they could not help with these issues, which are rather to be checked with the administrative offices.
To arrange an appointment, you can contact them by email:

Francesco Cecconi francesco.cecconi@unicatt.it
Matteo Bordi matteo.bordi@unicatt.it
Viviana Greco viviana.greco@unicatt.it
Emanuele Panatta emanuele.panatta@unicatt.it
Andrea Papait andrea.papait@unicatt.it
Eugenio Sangiorgi eugenio.sangiorgi@unicatt.it

8. 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 final score that needs to be achieved to pass the written test is 18. Passing the written test may give access to an oral examination (according to individual disciplines). During the oral examination, 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 at the written test.

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. PROGRAM

CELLULAR BIOLOGY 2 - MZ000063

Structure and content of the eukaryotic nucleus; chromatin and chromosomes; eukaryotic genome organisation. Basics of gene transmission and regulation in eukaryotic cells: DNA replication and repair; RNA transcription and post-transcriptional processing; genetic code; protein translation. Mechanisms of cellular sensing and communication: signal transduction and intracellular signalling, main signalling pathways and their implication in diseases.

CELLULAR BIOLOGY PRACTICALS - MZ000064

The practical sessions for Cellular Biology II focus on laboratory exercises designed to investigate gene expression in in vitro cell culture models. Techniques covered include:

1. Nucleic acids isolation;
2. cDNA synthesis: reverse transcription reaction;
3. Polymerase Chain Reaction (PCR).

BIOCHEMISTRY - MZ000061

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 - MZ000062

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

MOLECULAR BIOLOGY - MZ000065

Nucleotides and Nucleic Acids structures: Nucleic Acid Structure, Nucleic Acid Chemistry, Other Functions of Nucleotides; DNA and RNA Innovative Technology; DNA Cloning; The basics of controlling Gene Expression: From Genes to Genomes, From Genomes to Proteomes; Structure to function analysis: from gene to protein structures. Basics of Molecular Embryology: the paradigm of cell determination by time-space related gene expression; the Hox code; Introduction to the Molecular Biology of Cancer Cells; Introduction to Epigenetic regulation in Biomedicine.

HUMAN GENETICS - MZ000066

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.