GENE REGULATION AND CELL METABOLISM (A002229)

1. lingua insegnamento/language

English.

2. contenuti/course contents

Coordinatore/Coordinator: Prof. ALESSANDRO ARCOVITO
Anno di corso/Year Course: 1st
Semestre/Semester: 2nd
CFU/UFC: 11
Moduli e docenti incaricati /Modules and lecturers:
- BIOCHEMISTRY (A002305) - 5.5 CFU - SSD BIO/10 - Prof. Federica Iavarone, Alessandro Arcovito
- BIOCHEMISTRY PRACTICALS (A002306) - 0.5 CFU - SSD BIO/10 - Prof. Emanuele Panatta, Emanuele Panatta
- CELLULAR BIOLOGY 2 (A002307) - 2 CFU - SSD BIO/13 - Prof. Wanda Lattanzi
- CELLULAR BIOLOGY PRACTICALS (A002308) - 0.5 CFU - SSD BIO/13 - Prof. Valentina Saccone, Lorena Di Pietro, Federica Tiberio, Martina Sandona'
- HUMAN GENETICS (A002310) - 1.5 CFU - SSD MED/03 - Prof. Pietro Chiurazzi, Giuseppe Marangi
- MOLECULAR BIOLOGY (A002309) - 1 CFU - SSD BIO/11 - Prof. Francesco Cecconi

3. testi di riferimento/BIBLIOGRAPHY

CELLULAR BIOLOGY II
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. 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 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. prerequisiti/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. metodi didattici/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. altre informazioni/OTHER INFORMATION

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:

Alessandro Arcovito alessandro.arcovito@unicatt.it
Francesco Cecconi francesco.cecconi@unicatt.it
Federica Iavarone federica.iavarone@unicatt.it
Emanuele Panatta emanuele.panatta@unicatt.it
Pietro Chiurazzi pietro.chiurazzi@unicatt.it

Giuseppe Marangi giuseppe.marangi@unicatt.it
Wanda Lattanzi wanda.lattanzi@unicatt.it
Lorena Di Pietro lorena.dipietro@unicatt.it
Diego Sibilia diego.sibilia@unicatt.it
Federica Tiberio federica.tiberio@unicatt.it
Valentina Saccone valentina.saccone@unicatt.it

8. modalità di verifica dell'apprendimento/METHODS FOR VERIFYING LEARNING AND FOR EVALUATION

The evaluation process consists of **three steps**:
Step a) In-Progress Evaluation
During the semester, students will complete multiple-choice questionnaires and/or assignments during lectures. These will contribute to a **partial score**, broken down as follows:

- **Biochemistry**: up to **5 points**
- **Cell Biology**: up to **3 points**
- **Human Genetics**: up to **2 points**
- **Molecular Biology**: up to **2 points**

Step b) Written Test
A mandatory written test will include **50 multiple-choice questions**, divided into **two blocks**, to be completed in **90 minutes**:

- **BLOCK 1** – *Biochemistry*:
  - 26 questions
  - 46 minutes
- **BLOCK 2** – *Molecular Biology, Cell Biology II, and Human Genetics*:
  - 4 questions (Molecular Biology)
  - 12 questions (Cell Biology II)
  - 8 questions (Human Genetics)
  - Total: 24 questions in 44 minutes

Scoring Rules:

- 1 point per correct answer
- 0 points for incorrect or unanswered questions

Passing Criteria:

- A **minimum of 15 correct answers** is required **in each block** to pass.
- If **BLOCK 1** is failed, the student **cannot attempt BLOCK 2**.
- Failing either block requires repeating **both blocks** at the next exam session.

Final Score Calculation (after Step a and b)
To proceed without an oral exam, the student must obtain:

- **At least 18 points** from the **sum** of:
  - BLOCK 1 written score + in-progress evaluation of **Biochemistry**
- **At least 18 points** from the **sum** of:
  - BLOCK 2 written score + in-progress evaluation of **Cell Biology, Human Genetics, and Molecular Biology**

If both values 18, the **final grade** is the **average** of the two.
If either value < 18, proceed to **Step c (oral exam)**.
Step c) Oral Examination
The oral exam is **mandatory** for students who do **not** reach 18 in either component after steps a and b. It is **optional** for others.

- The student answers **4 questions**, each scored between **-1 and +1**
- Total oral score range: **-4 to +4**
- This score is **added** to the average from step b

Pass Condition: Final score after adding oral result must be **> 18**
Fail Condition: If the final score is still 18, the student must **repeat the entire exam**, starting from the written test of **both blocks**
Honors

- If the final average score (after oral, if taken) is **above 30**, the student will receive a final mark of:
  - **30/30 cum laude**

9. programma esteso/program

CELLULAR BIOLOGY II

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 II PRACTICALS:

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

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 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

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. Multifactorial traits. Cytogenetics: human chromosomes and karyotype; chromosome segregation 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.