POLI ESCOLA SUPERIOR SAÚDE TÉCNICO GUARDA	**SUBJECT DESCRIPTION**	**MODELO** PED.015.03

Course	**Pharmacy**					
Subject	**Molecular and Human Genetics**					
Academic year	2023/2024	*Curricular year*	3rd	*Study period*	1st semester	
Type of subject	Compulsory	*Student workload (H)*	Total: 135	Contact: 76,5	*ECTS*	5
Professor(s)	Ricardo Jorge Fernandes Marques					
☒ *Area/Group Coordinator* ☐ *Head of Department* *(select)*	Ricardo Jorge Fernandes Marques					

PLANNED SUBJECT DESCRIPTION

1. LEARNING OBJECTIVES

The curricular unit of Molecular and Human Genetics has the following objectives:

1.1. Integration of basic knowledge about the synthesis, structure and function of nucleic acids.

1.2. Identify how genes and genomes are organized at the molecular level.

1.3. Understand DNA replication, the molecules involved, the errors that can arise in the process and the mechanisms by which cells can repair them.

1.4 Understand the mechanisms of gene expression: transcription, RNA processing and translation.

1.5. Understand the rules of heredity (classical Mendelian genetics and its exceptions) and the integration of knowledge at the molecular level in the explanation of classical heredity.

1.6. Understand the function of the gene and the influence it exerts on the definition of the organism as a whole and the environment.

1.7. Understand the use of genetic techniques for the study and development of fundamental biological processes, in normal and pathological situations.

1.8. Recognize the bases of susceptibility to genetic diseases, as well as the methods involved in their study and diagnosis.

1.9. Understand the bases inherent to the understanding and application of the general concepts of Genetics in Pharmacogenomics, Biomedical Sciences and Gene Therapy.

1.9. Develop the ability to analyze scientific texts and explain the theoretical foundations of practical problems.

1.10. Develop the capacity for self-learning, research and choosing appropriate information.

2. PROGRAMME

2.1. DNA and biological information

2.2. DNA replication

2.3. Mechanisms of gene expression: transcription and RNA processing

2.4. The genetic code and protein synthesis

2.5. Regulation of gene expression in eukaryotes

2.6. The molecular basis of heredity and genetic diseases

2.7. Population genetics

2.8. Molecular organization of the human genome: identification of genes associated with diseases and pharmacological response

2.9. Implications for the development of new technologies derived from knowledge of the human genome

2.3. Practical syllabus

2.3.1. DNA quantification

2.3.2. Amplification of a DNA sequence by PCR

2.3.3. Agarose gel electrophoresis

3. COHERENCE BETWEEN PROGRAMME AND OBJECTIVES

The programmatic contents that make up the curricular unit are in agreement and allow achieving the objectives defined for this Curricular Unit. The integration of knowledge throughout the program is obtained through the analysis of scientific articles, research and choice of appropriate information as well as the necessary guidance developed by teachers for the self-learning process, which allows the student to acquire the skills necessary to develop their professional activity.

4. MAIN BIBLIOGRAPHY

Mandatory

• Genetics - A Conceptual Approach. Benjamin A. Pierce. (2016). 6th Edition. W. H. Freeman Publisher. ISBN-10:1319050964; ISBN-13: 978-1319050962.

• Regateiro, FJ (2003; several reprints until 2013). Handbook of Medical Genetics. Coimbra – University press. Coimbra.

• Supporting notes from the teacher.

Recommended

• Lewin's GENES XII. Jocelyn E. Krebs, Elliott S. Goldstein, Stephen T. Kilpatrick (2017). 12th Edition. Jones & Bartlett Learning. ISBN-13: 978-1284104493; ISBN-10: 1284104494.

5. TEACHING METHODOLOGIES (INCLUDING EVALUATION)

Continuous evaluation

The weight of the theoretical and theoretical-practical assessment in the final average will be 80% (40% 1st Test + 40% 2nd Test), the practical assessment will have a weight of 10% (written test) and the Seminar will have a weight of 10%.

Final evaluation

Failure to pass continuous assessment implies that an examination will be carried out, at the times provided for this purpose, of all syllabus content. The result of the exam evaluation, expressed on a scale of 0 to 20 values, will be the final classification of the curricular unit.

6. COHERENCE BETWEEN TEACHING METHODOLOGIES AND OBJECTIVES

The methodologies provided are consistent with the purpose of the curricular unit.

In theoretical classes, the expository method is encouraged by the application of the question and answer technique, carefully applied. Students will have access to all teaching material used (presentations, diagrams, images and videos) relating to each syllabus.

In theoretical-practical classes, autonomous work is encouraged through the systematic discussion of specific topics or problem solving.

The practical classes will be laboratory-based, aimed at learning the main techniques of molecular genetics. The students were stimulated by performing the techniques and discussing the results.

Tutorial guidance allows the teacher to work with students to guide and support their individual study.

7. ATTENDANCE

Success in this curricular unit (continuous assessment or final exam in the normal or resit period) requires participation and attendance, with a minimum mandatory attendance of 75% in theoretical-practical classes and 100% in practical laboratory classes. Students with Special Statutes and Conditions (e.g. student worker status) are governed by the benefits provided for in the legislation.

8. CONTACTS AND OFFICE HOURS

Contact: Ricardo Jorge Fernandes Marques, email: rmarques@ipg.pt, office 6.

Opening hours: Monday 2:30 pm – 4:30 pm

Tuesday 9:30-10:30; 1:30 pm – 2:30 pm

DATE

31 de outubro de 2023

SIGNATURES

Professor(s), Area/Group Coordinator or Head of Department signatures

Professor

(signature)

Area/Group Coordinator

(signature)