

Course Unit	Molecular Genetics and Pharmacogenomics	Field of study	-
Bachelor in	Pharmacy	School	School of Health
Academic Year	2023/2024	Year of study	2
Type	Semestral	Semester	1
Level	1-2	ECTS credits	5.0
Code	9549-803-2103-00-23		
Workload (hours)	135	Contact hours	T - TP 30 PL 30 TC - S - E - OT 7,5 O -

T - Lectures; TP - Lectures and problem-solving; PL - Problem-solving, project or laboratory; TC - Fieldwork; S - Seminar; E - Placement; OT - Tutorial; O - Other

Name(s) of lecturer(s) Maria Inês Pires Nogueiro

### Learning outcomes and competences

At the end of the course unit the learner is expected to be able to:

1. Knowing the genetic basis of heredity: DNA as the genetic material, its chemical nature and structural characteristics
2. Identify the structure and organization of the genome of different organisms (prokaryotes, eukaryotes and virus)
3. Knowing the molecular mechanisms of DNA replication, transcription and translation in prokaryotes and eukaryotes
4. knowing the laws of genetic inheritance and its exceptions
5. Describing and defining basic concepts pharmacogenomics and pharmacogenetics
6. Knowing the genes that which are largely responsible for variances in drug response and metabolism
7. Evaluate the types of polymorphism and its impact on the pharmacokinetics and pharmacodynamics
8. Acquire basic laboratory knowledge in the field of Molecular Genetics applied to Pharmacogenomics in the analysis of important polymorphisms in the treatment of cancer, CVD, etc.

### Prerequisites

Before the course unit the learner is expected to be able to:  
Not applied

### Course contents

Introduction to genetic bases. Laws of genetic inheritance and its exceptions. Eukaryotic, prokaryotes and virus genomes. The DNA replication, transcription and translation in prokaryotes and eukaryotes. Genetic variation and epigenetic mechanisms. Pharmacogenomics and Farmocogenetics: concepts. Genetic basis of drug response. Polymorphic variants leading to interindividual variability. Limitations and ethical issues.

### Course contents (extended version)

1. Introduction to genetics
  - Definition and evolution
  - Importance, applications and perspectives
2. Genetic transmission and the laws of genetic inheritance and its exceptions
  - Mendel's experiments and laws
  - Autosomal dominant and recessive inheritance
  - Multiple allelomorphism: ABO blood group
  - Sex-linked traits
  - X chromosome inactivation: The Lyon hypothesis. Barr body
  - Sex-limited and sex-influenced autosomal inheritance
3. DNA as the genetic material
  - Identification of DNA as genetic material
  - Chemical composition of DNA
  - DNA and RNA structure
4. Different types of genome: Eukaryotic, prokaryotes and virus genomes
  - Classification of viruses according to their genome
  - Bacterial genomes and plasmids
  - Mitochondrial genome
  - Nuclear genome
5. DNA replication in prokaryotes and eukaryotes
6. Transcription
  - Structure of RNA: rRNA, tRNA and mRNA
  - Steps of the transcription: initiation, elongation and termination
7. Translation and Genetic Code
  - Protein synthesis: stages, factors and enzymes
8. Genetic variation and epigenetic inheritance
  - Mechanisms in epigenetic regulation
  - Epigenetic modification by environmental factors
9. Pharmacogenomics and Pharmacogenetics: Personalized medicine.
  - Factors that contributed to the development of these new sciences.
  - Genetic Basis of Drug Response: (enzymes, carrier proteins and receptors).
10. Genetic polymorphisms leading to interindividual variability in drug response.
  - Available genotyping methods. Polymorphism analysis: Sequencing and qPCR protocols .
  - Commercial tests and interpretation of results: Oncology, Infectious Diseases and Coagulation.
11. Use of genomic information, for targeted drug development.
  - Recombinant DNA technology. Application in the production of new drugs.
  - Pharmacogenomics/Pharmacogenetics and their inclusion in clinical trials.
  - Ethical issues.

### Recommended reading

1. Allison L. A. (2012) Fundamentals of Molecular Biology, 2nd Edition, Wiley-Blackwell
2. Klug WS, Cummings MR, Spencer C, Palladino MA, 2015. Concepts of Genetics. 11th Edition. Pearson Education
3. Krebs J. E. , Goldstein E. S. , Kilpatrick S. T. (2013). Lewin's GENES XI, 11 ed. , Jones & Bartlett Learning

### Teaching and learning methods

Lectures using power point presentations. Lectures notes deposited in the e-learning resources. Laboratory classes.

**Assessment methods**

1. Alternative 1 - (Regular, Student Worker) (Final)
  - Intermediate Written Test - 40% (Practical evaluation)
  - Intermediate Written Test - 30% (Theoretical evaluation)
  - Final Written Exam - 30% (Theoretical evaluation.)
2. Alternative 2 - (Regular, Student Worker) (Supplementary, Special)
  - Final Written Exam - 40% (Practical evaluation)
  - Final Written Exam - 60% (Theoretical evaluation)

**Language of instruction**

Portuguese, with additional English support for foreign students.

**Electronic validation**

María Inês Pires Nogueiro	Carina de Fatima Rodrigues	Ana Maria Nunes Português Galvão	Adília Maria Pires da Silva Fernandes
15-01-2024	17-01-2024	18-01-2024	18-01-2024