Course Unit	Genetics			Field of study	Science Base		
Bachelor in	Biomedical Laboratory Sciences			School	School of Health		
Academic Year	2023/2024	Year of study	2	Level	1-2	ECTS credits	5.0
Type	Semestral	Semester	2	Code	9995-804-2202-00-23		

Workload (hours)	135	Contact hours	T -	TP 22,5	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. To recognize the genetic disease as an extreme of human variability;
2. To know the principal patterns of monogenic heredity;
3. Learn how to collect, register and interpret a family history To evaluate and calculate genetic risks in simple cases and determine the hereditary nature of a disease;
4. To identify the most important instruments of the molecular genetics and cytogenetic analysis in the genetic variability study. (polymorphism / mutation);
5. To know the molecular bases of genetic diseases;
6. To identify particular aspects of genetic diseases
7. Knowing extranuclear heredity
8. Understand how genes are distributed in the population (gene frequencies)

Prerequisites

Not applicable

Course contents

The material in this course encompasses diagnosis of genetic diseases, the study of inheritance of diseases in families, mapping of disease genes to their chromosome locations, the study of the molecular genetics of inherited disorders, provision of genetic counseling for families and investigations of methods for gene therapy.

Course contents (extended version)

1. The Human condition in Medical Genetics. Historical perspective and his impact in medicine.
2. Genetic transmission and genetic heredity laws and exceptions. Aspects of fenotypical expression.
   - Autosomal dominant and recessive characters
   - Multiple allelomorphism
   - Holandric genes and Sex-Linked Genes
   - X chromosome inactivation
   - Sex-limited and sex-influenced autosomal inheritance
   - Gene interaction. Epistasis
3. Molecular basis of hereditary
   - Nuclear and mitochondrial genome
   - Extranuclear inheritance
4. Genetic material alterations
   - Chromosomal mutations (classical cytogenetics and molecular cytogenetics)
   - Gene mutations
   - Classic Citogenetics and Molecular Citogenetics techniques.
   - Molecular biology techniques applied to genetic diagnosis.
   - Bioinformatics.
5. Genética Populacional:
   - Hardy - Weinberg Law (genotype and phenotype frequencies).
   - Factors affecting Hardy-Weinberg equilibrium.
   - Linkage desiquilibrium.
6. Prevention and treatment of genetic diseases:
   - Prenatal diagnosis;
   - Genetic counseilling: principles and techniques;
   - Genetic Therapie.
7. PRACTICAL LESSONS
   - Genetic Heredity exercices.
   - Population analysis: gene and genothype frequencies.
   - Recombiantion analysis annd genetic distance.
   - Molecular Biology Techniques applied to Genetic Diagnosis.
   - Several protocols for extracting DNA from whole blood.
   - PCR protocols and strategy for product analysis: restrition analysis, SSCA.
   - Sanger gene sequencing and fragments analysis in capillary electrophoresis.
   - Real time PCR: RNA analysis, gene expression of Beta globin
   - Classical Cytogenetics and Molecular cytogenetics: results analysis: FISH, MLPA

Recommended reading

1. Gelehrter, T. , Francis, C. & Ginsburg, D. (1998). Principles of Medical Genetics. USA: Lippincott Williams & Wilkins.
2. Steinberg, M. (2009). Disorders of Hemoglobin: Genetics, Pathophysiology, and Clinical Management. New York: Cambridge University Press.
3. Griffiths, A. , Gelbart, W. , Lewontin, R. & Miller, J. (2002). Modern Genetic Analysis integrating genes and genomes. USA: W. H. Freeman.
4. Strachan, T. & Read, A. (2004). Human Molecular Genetics. USA: Bios Scientific Publishers Ltd.
5. Passarge, E. , Borges-Osorio, M. R. , Robinson, W. R. (2004). Genetica texto e atlas. Porto Alegre: Porto Alegre Artmed.

Teaching and learning methods

The theoretical lessons will use the expository method and the students must solve practical exercise related to familial risk assessement. The course will include a practical component in which students carry out works on some of the themes, including the most current and relevant to the basic methods of DNA manipulation.

Assessment methods

1. Alternative 1 - (Regular, Student Worker) (Final)
   - Intermediate Written Test - 30% (The written exam covers the contents of theoretical lessons.)
   - Final Written Exam - 40% (The written exam covers the contents of practical lessons.)
   - Final Written Exam - 30% (The written exam covers the contents of theoretical lessons.)
2. Alternative 2 - (Regular, Student Worker) (Supplementary, Special)
   - Final Written Exam - 60% (The written exam covers the contents of theoretical lessons.)
   - Final Written Exam - 40% (The written exam covers the contents of practical lessons.)

Language of instruction

Portuguese

Electronic validation

Maria Inês Pires Nogueiro	Carina de Fatima Rodrigues	Ana Maria Nunes Português Galvão	Adília Maria Pires da Silva Fernandes
22-03-2024	04-04-2024	06-04-2024	07-04-2024