

## COURSE SYLLABUS

### **Genetics I**

**2425-1-H4102D002-H4102D008M**

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#### **Aims**

The course will provide the essential theoretical knowledge of biology and genetics, also focusing on the possible future application in the medical field. The subjects of the course will provide the necessary knowledge to understand the vital processes, both on the cellular and molecular level, as well as the laws of heredity and the processes involved in the generation of phenotypic diversity. The acquired knowledge will contribute to better understand the processes involved in normal and pathological situations.

#### **Contents**

molecular and cellular mechanisms responsible for gene expression and its regulation, analyzing epigenetic mechanisms, basic concepts of heredity and the transmission patterns of inherited traits; mechanisms which can generate phenotypic variants in men.

#### **Detailed program**

GENETICS – Human reproduction – Genetic variability – Inheritance – Genes: genotype and phenotype – Diploidy and reproduction. Homologous chromosomes, alleles and loci, homozygosity and heterozygosity – Mendel's laws – Alleles: wild-type, mutated and multiple ones, dominance and recessivity – Mendel's law's implementation: epistasis, penetrance and expressivity – Sex chromosomes. Sex determination – How to build and analyze a family tree – Chromosome X inactivation. Its implication in the phenotypic manifestations of genetic diseases – Test cross and inheritance of genes localized on different chromosomes – Crossing over and its genetic consequences – Recombination frequencies calculation and genetic maps – Principles and consequences of mitochondrial inheritance and genomic imprinting – Examples of monogenic inheritance: blood groups (ABO, Rh), color blindness – Multigenic inheritance and quantitative genetics – Characters showing a threshold effect – Multifactorial disorders –

Population genetics and Hardy-Weinberg equilibrium. CYTOGENETICS – Methods for chromosome analysis – Normal human caryotype – Chromosomal and genomic mutations and their effect on meiosis and phenotype – Deletions, inversions, duplications, translocations and non-disjunctions – Down's, Turner's and Klinefelter's syndrome – Chromosomal mutations and leukemia: Philadelphia chromosome and Burkitt's lymphoma – Germinal and somatic mutations, mosaicism. MOLECULAR GENETICS: Relationship between DNA content and organism complexity – DNA assembly in the nucleus of eukaryotic cells – Structural differences between prokaryotic and eukaryotic genes – Genome organization in prokaryotic and eukaryotic cells. Characteristics of human genome – Gene mutations: development mechanisms – Mutation consequences on gene products – Examples of autosomic dominant and recessive mutations, as well as X-linked ones – Mitochondrial gene mutations – Genomic instability - DNA polymorphisms and their use as genetic markers – Elements of developmental biology – Immunogenetics. Generation of antibody diversity - The human genome project: future implications -Elements of gene therapy

## **Prerequisites**

Basic sciences (chemistry, physics)

## **Teaching form**

22 two-hour classes delivered in presence.

3 two-hour classes with an initial part delivered by the professor aiming to generate an interactive work by the students.

All classes will require attendance in person.

## **Textbook and teaching resource**

Thompson & Thompson Genetics and Genomics in Medicine, 9e, 2023; Peter Russell iGenetics: A Molecular Approach. Pearson, 2016

## **Semester**

Second semester

## **Assessment method**

These subjects will be evaluated within the exam of the integrated course, as described in the Syllabus of the entire course.

## **Office hours**

by appointment

## **Sustainable Development Goals**

GOOD HEALTH AND WELL-BEING | QUALITY EDUCATION | GENDER EQUALITY

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