Universitat de Lleida

DEGREE CURRICULUM

GENETICS

Coordination: FIBLA PALAZON, JUAN

Academic year 2023-24

GENETICS 2023-24

Subject's general information

Subject name	GENETICS
Code	101504
Semester	1st Q(SEMESTER) CONTINUED EVALUATION
Typology	

Degree	Course	Character	Modality
Bachelor's Degree in Biomedical Sciences	2	COMMON/CORE	Attendance-based
Master's Degree in Biomedical Research		COMPLEMENTARY TRAINING	Attendance-based

Course number of credits (ECTS)	6
Type of activity, credits, and groups	

Activity type	PRALAB	PRAULA	TEORIA
Number of credits	1	1	4
Number of groups	3	2	1

Coordination	FIBLA PALAZON, JUAN
Department	BASIC MEDICAL SCIENCES
Important information on data processing	Consult this link for more information.

GENETICS 2023-24

Teaching staff	E-mail addresses	Credits taught by teacher	Office and hour of attention
FIBLA PALAZON, JUAN	joan.fibla@udl.cat	6	
FIBLA PALAZON, JUAN	joan.fibla@udl.cat	0	
LOPEZ ORTEGA, RICARDO ENRIQUE	ricard.lopez@udl.cat	3	

Competences

CE1. Apply and formulate mathematical and physical concepts of relevance for the study of human biology.

CE2. Apply the mathematical and physical concepts learned in biomedical experiments and research.

CE11. Describe the mechanisms of storage and processing of genetic information, as well as the different levels of organization of the human genome.

CE12. Apply the basics of genetics, as well as methods for the study of genetic pathologies

CE18. Critically evaluate and use clinical and biomedical information technologies and sources to obtain, organize, interpret and communicate clinical, scientific and health information

CE19. Use laboratory material and apply basic laboratory techniques.

Subject contents

Module I: Genetic analysis of the phenotype (12 hours)

- UNIT 1. Introduction to Genetics and Heredity- Objectives and scope of Genetics. Structure of hereditary material, inheritance and variation. Genotype, Phenotype and Environment. Basic nomenclature in genetics. Genetic analysis of the phenotype. Chromosomal basis of inheritance: Mitosis and meiosis. Chromosomes and the cell cycle. Behavior of chromosomes during Mitosis and Meiosis. Genetic consequences of Meiosis.
- UNIT 2. Introduction to the genetic analysis of the phenotype. Type of phenotypes. Genotype-environment interaction. Monogenic, polygenic and multifactorial characters. Concept of heritability. Empirical determination of the heritability of a character. Determination of the genetic basis of a character. Genetic analysis of complex characters. Twin studies: concordance and correlation of character in related individuals. Multifactorial characters. Quantitative traits. Phenotypic distribution of a quantitative character. Analysis of variance. Model "threshold". Directed selection experiments. Response to selection.
- UNIT 3. Genetic analysis of monogenic characters. Mendelian inheritance models. Segregation models: monohybridism / dihybridism. Allelic relationships: dominance and recessivity.
- UNIT 4. Inheritance of genes linked to sex chromosomes. Genetic determination of sex. Environmental factors and sex determination. Models of sexual determination: chromosomal balance and homo-heterogametic sex. Dose compensation. Comparative structure of the sex chromosomes. Inactivation of the X chromosome. Heredity influenced by sex. Inheritance limited to one sex.
- UNIT 5. Extrachromosomal inheritance patterns. Genome of cytoplasmic organelles and symbionts. Organization of the mitochondria genome. Organization of the chloroplast genome. Characters with maternal effect.
- UNIT 6. Extensions of the Mendelian analysis. Genetic analysis of linked genes. Ligation and recombination. Alteration of phenotypic proportions in linked genes. Recombination frequency and genetic distance Genotypic interactions. Epistasia. Allelic incompatibility and lethality. Penetration and expressiveness. Pleiotropia. Epigenetic factors: Genetic footprint

Module II. Genetic variability (12 hours)

- UNIT 1. Mechanisms of genetic change - Mutation: origin and types. Spontaneous mutation and induced mutation. Chromosomal mutations. Karyotype. Numerical and structural changes of chromosomes. Aneuploids in humans. Ploidy in Plants. Gene mutations. Types of gene mutations. Recombination not homologous. Phenotypic effects of mutations.
- UNIT 2. Study and characterization of genetic variability - Concept of genetic polymorphism. Study of polymorphism at the DNA level. Type of polymorphic DNA variations: SNPs, insertions / deletions and repeats of variable number. Genotyping methods and techniques. Applications of genetic polymorphisms.
- UNIT 3. Genome mapping - Physical maps. Sequence positioning using probes. Somatic hybridization. Other physical mapping techniques. Genetic maps. Recombination frequency and linkage maps. Use of genetic polymorphisms in mapping. Genetic maps in the human species. Lod score method. Module III.

Genetic analysis of the population (6 hours)

- UNIT 1. Population genetics - Mendelian population concept. Phenotypic, genotypic and allelic frequencies. Hardy-Weinberg equilibrium. Hardy-Weinberg equilibrium distortions. Consanguinity and heterosis. Change of allelic frequencies. Mutational equilibrium, Migration, Selection: biological efficacy. Stochastic changes: Genetic drift. Population size: founder effect and bottlenecks. (4 hours).
- UNIT 2. Evolutionary medicine - Evolutionary vision of the health-disease bonus. Obesity, an evolutionary perspective. Lactose intolerance. Cancer: a Darwinian view.
- UNIT 3 . Personalized Medicine - Genetic Determinism vs. susceptibility. The concept of penetrance. Massive genetic analysis technologies. Personalized medicine in cancer treatment. Personalized Medicine in the prescription of a treatment. Pharmacogenetics.

Problem sessions

- Meiosis and biological cycles.
- Mendelian proportions and inheritance patterns
- Applications of genetic polymorphisms: genetic identification and study of paternity. Study of linkage in human families.
- Calculation of phenotypic, genotypic and allelic frequencies. Checking the Hardy-Weinberg Equilibrium. Calculation of genotype frequencies assuming Hardy-Weinberg. Association studies

Practical activities Sessions

- Laboratory of practices
  - LABORATORY PRACTICE - Study and characterization of the human karyotype.

- Computer classroom sessions
  - INFORMATICS 1 - Mendelian inheritance patterns - Simulation crosses (I)
  - INFORMATICA 2 - Mendelian inheritance patterns - Simulation crosses (II)

Methodology

Tipus d'Actividad	Descripció resumida de l'actividad
TEO	Master classes of theoretical content
LAB	Students must obtain viable peripheral blood cells in order to characterize the chromosomal complement and thus obtain the karyotype. An optical microscope observation of the processed material and pathological samples will be carried out in order to study the different types of anomalies observed.
INF	On-line applications to analyze the human genome.
PRO	Resolutions of the proposed problems

Evaluation

Assessment of learning Type of evaluation

- Theory 70% (Written test on contents theoretical classes, problems and laboratory practices)
- Practices 15% (Solving bioinformatics problems)
- Problems 15% (Problem solving in class)

Bibliography

Basic bibliography

- Jorde L y cols (2006). Genética Médica. Elsevier
- Sudbery P. (2004). Genética molecular humana. Pearson / Prentice Hall.
- Novo Villaverde FJ. (2007). Genética humana: Conceptos, mecanismos y aplicaciones de la Genética en el campo de la Biomedicina. Pearson / Prentice Hall
- Nussbaum RL, et al. (2004). Genetics in Medicine. Thompson & Thompson.
- Solari AJ. (2004). Genética Humana, Fundamentos y aplicaciones en Medicina. Editorial Médica Panamericana

Complementary bibliography

- Griffiths, Miller, Lewontin & Suzuki. Genetica. McGraw-Hill / Interamericana de Espana, SA WS Klug, MR Cummings, Genetica (Pearson Educación, SA, ed. 1 ª, 1998).
- Anthony JF Griffiths, Jeffrey H. Miller, David T. Suzuki, Richard C. Lewontin, William M. Gelbart, An Introduction to Genetic Analysis (WH Freeman & Company, ed. 8th, 2004).
- MRC William S. Klug, Essentials of Genetics (Prentice Hall, ed. 5th, 2004).
- DPSEJ Gardner, MJ Simmons, Principles of Genetics (John Wiley and Sons Ltd, ed. 8th, 1991). RH Tamarin, Principles of Genetics (William C Brown Pub, ed. 6th, 1999).
- EJ Eisen, The Mouse in Animal Genetics And Breeding Research (World Scientific Publishing Company, 2005).
- JH Gillespie, Population Genetics: A Concise Guide (Johns Hopkins University Press, ed. 2nd, 2004)