COURSE DATA

DATA SUBJECT

Code: 33136
Name: Genetics and cytogenetics
Cycle: Undergraduate Studies
ECTS Credits: 9
Academic year: 2025-26

STUDY (S)

Degree	Center	Acad. year	Period
1109 - Degree in Biochemistry and Biomedical Sciences	Facultat de Ciències Biològiques	2	Annual

SUBJECT-MATTER

Degree	Subject-matter	Character
1109 - Degree in Biochemistry and Biomedical Sciences	Genética y biología molecular	COMPULSORY

COORDINATION

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SUMMARY

The subject of genetics and cytogenetics is offered in the second year of Bachelor's degree in Biochemistry and biomedical CC (2009 Plan). It is a compulsory subject, together with the genomics, genetics of development, human genetics, technical subjects of genetic analysis and genetic engineering aims to provide the student the basics relating to biological inheritance as well as the conceptual and methodological tools that enable it to carry out, in their professional work, tasks related to genetic analysis and the clinical genetics.

The student studied this subject at the same time to subject structure of macromolecules, methods in biochemistry, biosynthesis of macromolecules, etc., which will complement the skills and basic knowledge related to the foundations of the molecular biology and cell, in particular on the structure of nucleic acids, their replication, transcription and translation, cell cycle, and phenomena of importance from the genetic perspective such as mitosis and meiosis, the two mechanisms responsible for the transmission of hereditary information.

PREVIOUS KNOWLEDGE

RELATIONSHIP TO OTHER SUBJECTS OF THE SAME DEGREE

There are no specified enrollment restrictions with other subjects of the curriculum.

OTHER REQUIREMENTS

COMPETENCES / LEARNING OUTCOMES

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Adquirir conocimientos teóricos sobre la estructura, función y evolución de los genomas.

Capacidad de aprendizaje autónomo.

Capacidad de comunicación oral y escrita.

Capacidad de manejar el inglés como lengua extranjera.

Capacidad de resolución de problemas.

Capacidad de utilizar las nuevas tecnologías de información y comunicación.

Capacidad para el trabajo en equipo y la cooperación.

Capacidad para trabajar en el laboratorio de genética y biología molecular incluyendo seguridad, manipulación, eliminación de residuos y registro anotado de actividades.

Comprensión de la lógica molecular de los seres vivos como producto de la evolución.

Conocer las aplicaciones de los conocimientos en genética y biología molecular en el diagnóstico de enfermedades humanas.

Conocer los elementos comunes y los diversos de la genética y la biología molecular de los diferentes tipos de organismos vivos.

Conocer y comprender las bases moleculares de la información genética y los mecanismos de su transmisión y variación.

Desarrollo de habilidades para comprender metodología e interpretar resultados científicos.

Desarrollo de la capacidad de razonar y aplicar el método científico.

Relacionar las características estructurales y funcionales de las macromoléculas.

Skills in analysis and synthesis.

Tener una visión integrada de las respuestas celulares a los efectores y cambios ambientales.

Tener una visión integrada del metabolismo celular y la expresión génica relacionándolas con los distintos compartimentos celulares.

DESCRIPTION OF CONTENTS

1. Unit 1. Introduction. Basic concepts.

Genetics. Gene, locus, allele. Phenotype vs Genotype. Development Noise. Reaction Standard. Genetic Analysis. Mutation and types. Relationships between alleles. Symbology

2. Unit 2. Patterns of inheritance

A little history. Mendel's experiments. Experimental methodology in Mendel's works. The monohybrid cross: principle of segregation. Verification of the principle of segregation. The dihybrid crossing: principle of independent transmission. Extension to the polyhybrid cross.

3. Unit 3. Calculation of proportions and hypothesis testing.

Predict the proportions of the offspring in a cross. Basic concepts of probability. Product rule. Addition rule. Double entry tables and branching diagrams. Development of the binomial. Application of the binomial in trihybridism / polyhybridism. Use of the binomial in families. Hypothesis contrast. Statistical tests: Chi-square.

4. Unit 4. Chromosomal theory of heredity.

Suton and Boveri works. Chromosomal theory of heredity. Genes and meiosis. Cytological basis of segregation. Morgan and Bridges experiments. Meiotic nondisjunction. Chiasmas (Janssen). Crossover. The first genetic map. Cytological evidence of crossover. Creighton and McClintock experiments

5. Unit 5. Extensions of the Mendelian analysis.

Relationships between alleles of a gene. Multiple allelism. Dominance series between alleles. Complementation test. Alleles of self-sterility in plants. Blood groups. Lethal alleles. Inheritance linked to sex chromosomes. Interaction between genes. Genes and metabolism. Epistasia. Influence of the environment. Penetrance. Expressiveness Maternal effect. Age. Sex. Phenocopies. Cytoplasmic inheritance. Mitochondrial diseases.

6. Unit 6. Linkage, recombination and genetic maps in eukaryotes.

Transmission of linked genes. Terminology in linkage crosses. Complete ligation. Recombination and Linked Genes. Linkage detection. Coupling and repulsion. Two-point crossover. Calculation of distances. Distance in a dihybrid crossing. Chi-square test. The three point map. Order of genes. Calculation of distances. Contingency test. Limits of linkage maps. Accurate calculation of distances. Map function. Interference and Coincidence. Corrections. Genetic maps versus physical maps. Importance of recombination maps. Mitotic recombination.

7. Unit 7. Genetics of quantitative traits.

Qualitative vs quantitative hereditary characters. Johannsen experiences. Nilsson-Ehle experiences. Polygenes or multiple factors. Edward East experiences. Quantitative characters. QTLs: examples and applications. Influence of the environment and threshold effect. Basic statistical analysis applied to quantitative characters. Heritability, examples and calculation.

8. Unit 8. The metaphase chromosome.

The chromatin fiber: euchromatin and heterochromatin. Lateral and longitudinal differentiation of the metaphase chromosome. Centromere Telomeres. Chromomers. NOR region.

9. Unit 9. The karyotype.

Shape, size and number of chromosomes. The human karyotype. Chromosomal banding: types of banding. Other chromosome identification techniques. Accessory chromosomes. Polygenic chromosomes.

10. Unit 10. Sex chromosomes.

Chromosomal determination of sex. Cytogenetic properties of sex chromosomes. Sex chromatin and gene dose compensation. Molecular mechanism of X chromosome inactivation. Evolution of sex chromosomes in vertebrates. Genetic alterations of sex determination in humans.

11. Unit 11. Structural chromosomal variations I.

Deletions: types, origin, genetic and cytogenetic consequences. Deletions in human chromosomes. Chromosomal rings Duplications: types, origin, genetic and cytogenetic consequences. Duplications in human chromosomes. Isochromosomes. Microdeletion and duplication syndromes

12. Unit 12. Structural chromosomal variations II.

Investments: types, origin, genetic consequences. Cytogenetic behavior of structural heterozygotes. Translocations: origin and types. Robertsonian translocations: centric fusion. Multiple translocations.

13. Unit 13. Numerical chromosomal variations I.

Cell cycle variations and their consequences. Haploidy. Haploidy in angiosperms: origin, identification, cytogenetic behavior and applications in genetic improvement. Polyploidy: origin and identification. Cytogenetic behavior of polyploids. Artificial polyploidy and genetic improvement. Polyploidy in humans. Uniparental diploidy

14. Unit 14. Numerical chromosomal variations II.

The failures in meiotic disjunction and their consequences. Aneuploidy: definition and types. Origin and cytogenetic behavior of aneuploids. Human autosomal aneuploidies and associated syndromes. Down Syndrome. Aneuploidies of human sex chromosomes and associated syndromes. Mixoploids: mosaics and chimeras. Uniparental dysomy.

15. Problems

Prediction of offspring
Exceptions
Pedigree analysis
Linkage, recombination and genetic distance
Structural variations
Numerical variations

16. Practice

PRACTICE 1. Segregation of Characters. Observation of independent segregation against segregation linked genes. Application of statistical analysis of experimental results. Estimating the distance between linked genes. (8.5 hours)

PRACTICE 2. Polytene chromosomes. Preparation and observation of polytene chromosomes. Detection and study of chromosomal inversions. (6.5 hours)

WORKLOAD

PRESENCIAL ACTIVITIES

Activity	Hours
Theory	53,00
Laboratory	14,00
Classroom practices	23,00
Total hours	**90,00**

NON PRESENCIAL ACTIVITIES

Activity	Hours
Attendance at other activities	0,00
Individual or group project	20,00
Independent study and work	87,00
Preparation of lessons	28,00
Preparation for assessment activities	0,00
Resolution of case studies	0,00
Total hours	**135,00**

TEACHING METHODOLOGY

Class Theory: A total of 42 sessions are needed one hour to cover this facet teacher. The teacher will present the most relevant content for the course, using the media necessary for agile development and consistent application of them. Classes of problems: it held 23 sessions of one hour throughout the course, coordinated with the lectures, usually at the end of each of the sections of the agenda. The teacher will prepare a series of problems for each topic or subject block that will work individually (through personal preparation thereof) and collectively (through presentation and discussion of them in group class) various aspects Hands-on labs: They are of compulsory attendance. There will be 7 sessions of 2 hours each. Students should come equipped with a lab coat and safety glasses.
The activity will involve the preparation and presentation of a seminar, lasting approximately 30 minutes for the students in groups of three and active participation in the discussion of all the seminars. In \\"Genetics and Cytogenetics\\" will be held 5 seminars. The seminar activities will be binding.

EVALUATION

First Call

The concepts worked in the theoretical sessions, as well as the student's ability to face and solve genetic problems, will be evaluated continuously throughout the course, as far as possible, by carrying out various activities including questionnaires (kahoot or moodle type), designing and solving genetic problems in the classroom, attendance at specific face-to-face or "online" conferences and questionnaires related to them and the completion of two written tests at the end of each semester.

The value of the written tests carried out at the end of the first and second semester will be 85% of the grade for the subject. Continuous assessment activities may add up to an extra 10% on the grade for the course.

Laboratory: Attendance is mandatory. The f laboratory sessions and the analysis of results vill be evaluated. The value of the laboratory grade will be 10% of the total.

Seminars: The completion of this activity is mandatory. It will assess the capacity for synthesis and integration of information from participating students, the clarity and quality of exposition and defense of the questions made by students and teachers. The value of the seminar grade represents 5% of the total.

The final grade for the course will be the sum of the different sections: Theory / problems, laboratory practices and seminars, as long as a minimum of 5 is achieved in each of them. In addition, as long as the grade for the subject exceeds 5, up to an extra 10% can be achieved with the continuous assessment activities planned during the course. In the case of the Theory / problems section, the average of the tests written at the end of each semester must reach at least 5. If in one of the two tests the 5 is not reached, it

can be compensated from 4 with the other written test.

Second call: The grade obtained in the first or second semester will be saved for the second call, if it is greater than 5 out of 10, unless the student waives this (performing and presenting the corresponding section of the review of the second call).The grade obtained in the laboratory practices and seminars is unique and will be saved during a course if necessary.

Note from the Department of Genetics, \"students are reminded that the waiver is not possible to score on the subject, once published, both in the evaluation of participation in classroom teaching activities (laboratory, problems, seminars, etc. . .) the different tests and evaluation of documents submitted for this assessment (tests, reports, etc..)\".

REFERENCES

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- Nussbaum, R.L.; McInnes, R.R. and Willard, H.F. (2016). Thompson & Thompson Genética en Medicina. 8ª Edición. Ed. Elsevier Masson. ISBN: 9788445826423 Pascual, L i Silva, F. (2018). Principios básicos de genètica. 1ª edició. Ed. Síntesis. ISBN9788491711063 Pierce, B (2015) Genética: Un enfoque conceptual Panamericana ISBN-10: 8498353920 Problemas Benito, C. 141 Problemas de Genética. (2015). 1ª edició. Ed. Síntesis. ISBN 9788490772195 Pierce, B.(2023) Fundamentos de Genética. Conceptos y Relaciones. Panamericana, ISBN 10: 950060275X