UAB
Universitat Autònoma de Barcelona

Human Genetics

Code: 106687
ECTS Credits: 5

2024/2025

Degree	Type	Year
2502442 Medicine	OB	2

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Teachers

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Teaching groups languages

You can view this information at the end of this document.

Prerequisites

A good knowledge of Catalan and Spanish is indispensable, vehicular languages in which the classes will take place.

It is advisable that the students have a good knowledge of English, since many of the information sources of this subject are in this language.

It is convenient that the student has achieved basic skills in Cell Biology, Biochemistry and Molecular Biology.

It is convenient that the student knows the basic principles of genetics.

Objectives and Contextualisation

The subject is scheduled in the second year of the Medicine degree. Its general objective is to give students all the necessary information that will allow them to acquire knowledge about the organization, function and regulation of genes in normal conditions and will enable them to understand the mechanisms involved in genetic-based diseases.

The student will acquire advanced knowledge about human genome; epigenetics and regulation of gene expression; mutation and repair of DNA; pharmacogenomics; forensic genetics; genetics of development; inheritance patterns; cytogenetics; rare diseases; cancer genetics and population genetics.

Competences

- Communicate clearly, orally and in writing, with other professionals and the media.
- Critically assess and use clinical and biomedical information sources to obtain, organise, interpret and present information on science and health.
- Demonstrate basic research skills.
- Demonstrate understanding of the importance and the limitations of scientific thought to the study, prevention and management of diseases.
- Demonstrate understanding of the mechanisms of alterations to the structure and function of the systems of the organism in illness.
- Demonstrate understanding of the organisation and functions of the genome, the mechanisms of transmission and expression of genetic information and the molecular and cellular bases of genetic analysis.
- Demonstrate, in professional activity, a perspective that is critical, creative and research-oriented.
- Formulate hypotheses and compile and critically assess information for problem-solving, using the scientific method.
- Indicate the basic diagnosis techniques and procedures and analyse and interpret the results so as to better pinpoint the nature of the problems.
- Recognize the determinants of population health, both genetic and dependent on gender, lifestyle, and demographic, environmental, social, economic, psychological and cultural factors.

Learning Outcomes

1. Apply the basic techniques used habitually in the genetics laboratory.
2. Communicate clearly, orally and in writing, with other professionals and the media.
3. Compare the techniques and methods that help in genetic diagnosis.
4. Demonstrate basic research skills.
5. Demonstrate, in professional activity, a perspective that is critical, creative and research-oriented.
6. Describe the anomalies of human chromosomes and evaluate their consequences.
7. Describe the molecular bases of DNA mutation and repair.
8. Describe the organisation, evolution, inter-individual variation and expression of the human genome.
9. Explain the importance of research in the field of genetics.
10. Explain the transmission mechanisms of genetic material.
11. Formulate hypotheses and compile and critically assess information for problem-solving, using the scientific method.
12. Identify the concepts and language of genetics and consult the scientific literature in the area of human genetics.
13. Identify the distribution of genetic diseases in a given population taking their origin into account.
14. Identify the epigenetic factors involved in the control of gene expression.
15. Identify the genetic bases for the main diseases with a genetic basis or component.
16. Identify the genetic bases of human development.
17. Interpret scientific publications, and solve problems and case studies in the area of genetics.
18. Interpret the results of a scientific project.
19. Relate the genetic dysfunction with pathological phenotype.
20. Understand scientific texts and write review papers on human genetics and genetic diseases.

Content

Contents of THEORY (by topics):

I. Human genome
1. Human genome I
2. Human genome II

II. Cytogenetics
3. The normal human chromosome
4. Numerical chromosomal abnormalities
5. Unbalanced structural chromosomal alterations
6. Balanced structural chromosomal alterations

III. Inheritance patterns
7. Autosomal inheritance
8. Sex-linked inheritance
9. Multifactorial and polygenic inheritance
10. Mitochondrial inheritance

IV. Gene expression
11. Control of gene expression
12. Epigenetics
13. Variability in gene expression

V. Mutation and DNA repair
14. Molecular bases of the mutation
15. Nucleotide expansion mutations
16. Mechanisms of DNA repair
17. Pharmacogenomics

VI. Normal developmental and dysfunctional genetics
18. Genomic imprinting
19. Microduplication / deletion syndromes. Rare diseases
20. Prenatal and pre-implantation diagnosis
21. Genetics of metabolic and endocrine disorders
22. Genetic bases of mental disorders
23. Genes of control of embryonic development

VII. Cancer genetics and genomics
24. Cancer genetics I
25. Cancer genetics II

Detailed distributive blocks by topic:

I-1. Human genome I: general characteristics, protein coding genes, non-coding RNA genes, splicing, genome transcription.
I-2. Human genome II: repetitive elements, regulatory elements, genome variability.

II-3. The normal human chromosome: chromosome structure, centromeres, telomeres, chromosomel identification, karyotype, mechansims of chromosomal segregation
II-4. Numerical chromosomal anomalies: polyploidies; aneuploidies: origin and consequences; mosaics,trisomies and viable monosomies in the human species, molecular bases of the Down and Turner syndromes.
II-5. Unbalanced structural chromosomal alterations: origin, deletions, duplications, ring chromosomes, isochromosomes, phenotypic effects, nomenclature.
II-6. Balanced structural chromosomal alterations: pericentric and paracentric inversions: origin, risk of anomalies in offspring; reciprocal translocations: origin, balanced carriers and risk of anomalies in offspring; Robertsonian translocations: origin, balanced carriers and risk of anomalies in offspring; phenotype of balanced structural anomalies.

III-7. Autosomal inheritance: detection of genetic diseases in medical practice, characteristics and pattern of transmission of autosomal dominant inheritance, characteristics and transmission pattern of autosomal recessive inheritance, detection of heterozygotes in the population.
III-8. Heredity linked to sex: inheritance linked to the recessive and dominant X chromosome, inheritance linked to the Y chromosome.
III-9. Multifactorial inheritance: heritability, search for candidate genes, genetic and environmental basis, normal characters of continuous variability, multifactor alterations with threshold, common diseases that affect the adult population.
III-10. Mitochondrial inheritance: mitochondrial DNA, characteristics of mitochondrial inheritance, pattern of transmission of mitochondrial alterations, mitochondrial diseases.

IV-11. Gene expression: mechanisms of control and regulation of gene expression, microRNA and lncRNA, RNA editing.
IV-12. Epigenetics: epigenetic factors, modification of DNA, modificationof histones, inactivation of chromosome X.
IV-13. Variability in gene expression: genotype-phenotype relationships, multiple allelomorphism, phenotype of heterozygotes, reduced penetrance, variable expressivity, pleiotropy, heterogeneous.

V-14. Molecular bases of the mutation: concept and types of mutations, sequence mutations, structural mutations, chromosomal mutations, nomenclature of mutations, mutagenic agents.
V-15. Mutations by nucleotide expansion: molecular mechanisms, trinucleotide expansion, examples of associated diseases.
V-16. Mechanisms of DNA repair: cellular response to genetic damage, main mechanisms of DNA repair, diseases associated with errors in DNA repair.
V-17. Pharmacogenomics: response to drugs, polymorphisms of metabolizing molecules, transporters and drug receptors, pharmacological targets.

VI-18. Genomic imprint: concept, genes and imprinted chromosomal regions, alterations influenced by imprinting.
VI-19. Genetic bases of microduplication / deletion syndromesand rare diseases: definition and characteristics, examples, genetic counseling, genetic analysis.
VI-20. Prenatal and pre-implantation diagnosis: prenatal and pre-implantation diagnostic techniques, clinical guidelines, example cases.
VI-21. Genetics of metabolic and endocrine disorders: molecular alterations in the development and function of the metabolic and endocrine system. Types of diabetes.
VI-22. Genetic basis of mental disorders: genetic origin, Parkinson's disease, tools for early detection, genetic basis of behavioral disorders, Alzheimer's disease
VI-23. Genes of control of embryonic development: general characteristics, transcription factors and signal molecules, HOX genes.

VII-24. Cancer genetics I: oncogenes and tumor suppressor genes, types of cancer, accumulation of somatic mutations in the tumor cell, genomic alterations and cancer.
VII-25. Cancer genetics II: carcinogenesis models, solid tumors, hematological neoplasms.

Contents of SEMINARS:

Seminar 1: Fundamentals of DNA technologies
Seminar 2: Gene therapy
Seminar 3: Genetic identification and forensic genetics
Seminar 4: Population genetics
Seminar 5: Application of genetics in the clinical laboratory
Seminar 6: Genetic counseling
Seminar 7: Hereditary cancer syndromes

Contents of LABORATORY PRACTICES:

PLAB1: Analysis of the normal and altered human karyotype
PLAB2: Identification of polymorphisms by using molecular genetic techniques

PLAB3: Interpretation of sequence and structural variants of the genome

Activities and Methodology

Title	Hours	ECTS	Learning Outcomes
Type: Directed			
CLASSROOM PRACTICES (PAUL)	4	0.16	1, 20, 2, 4, 5, 6, 10, 11, 12, 13, 15, 17
LABORATORY PRACTICES (PLAB)	11	0.44	1, 3, 4, 5, 12, 15
SEMINARS (SEM)	7	0.28	1, 3, 8, 6, 10, 9, 11, 13, 15, 17
THEORY (TE)	25	1	1, 3, 8, 6, 7, 10, 9, 14, 13, 15, 16
Type: Supervised			
TUTORIES	14	0.56	1, 20, 2, 3, 4, 5, 8, 6, 7, 10, 11, 12, 14, 15, 17
Type: Autonomous			
READING ARTICLES / REPORTS OF INTEREST	22	0.88	1, 9, 11, 17
READING ARTICLES /REPORTS OF INTEREST	22	0.88	1, 6, 9, 13, 17
WORK PREPARATION	12	0.48	4, 9, 11, 17

Theoretical classes: 25 sessions. Systematized exposition of the syllabus of the subject, giving relevance to the most important concepts. The students acquire the basic scientific knowledge of the subject in theory classes, which will complement the personal study of the topics discussed and with problem-based learning methodology. Students can find a summary of the material used in class in the Virtual Campus and / or Moodle before or after the lecture.

Seminars: 7 sessions. Exhibition, in small groups, of relevant subjects of the subject and clinical cases. This methodology will allow students to review the most important or most basic topics necessary for understanding the subject.

Classroom practices (problems): 4 sessions. Exposition and resolution of cases and genetic problems presented by the professor.

Laboratory practices: 3 sessions. Exposure and application of the different techniques used in basic and molecular cytogenetics, interpretation of structural and polumorphic variants, and their clinical applicability.

IMPORTANT NOTE: Prior to the completion of laboratory practices, students must have completed the test that certifies the knowledge of the contents of the risk prevention manual and upload it to the Virtual Campus and / or Moodle. They are essential requirements to perform practices 1 and 2 take a lab coat and show the teacher a signed copy of the risk prevention test.

IMPORTANT NOTE: *The proposed teaching methodology may undergo some modification depending on the attendance in person restrictions.*

Annotation: Within the schedule set by the centre or degree programme, 15 minutes of one class will be reserved for students to evaluate their lecturers and their courses or modules through questionnaires.

Assessment

Continous Assessment Activities

Title	Weighting	Hours	ECTS	Learning Outcomes
Lecture: Written assessments through objective tests: multiple choice items	70%	6	0.24	1, 2, 3, 8, 6, 7, 10, 12, 14, 13, 15, 16, 19
Practice: written assessments through objective tests: Practical cases solving	20%	1	0.04	1, 20, 2, 3, 4, 5, 10, 9, 11, 12, 15, 18, 17
Problems: written evaluations through objective tests: Problem solving	10%	1	0.04	3, 10, 14, 13, 15

Evaluation

A. The competences acquired in theory classes, seminars and classroom practices (or genetic problems) of this subject will be evaluated as follows:

1. Continuous evaluation: it will be divided into two partial exams:

First partial:

• Multiple choice objective test of the knowledge acquired in theory classes (topics 1-13) and seminars 1-3. This test must be passed with a grade of 4,8 or higher to be used for the average. This test corresponds to 35% of the final grade of the subject.

Second partial:

• Multiple choice objective test of the knowledge acquired in theory classes (topics 14-25) and seminars 4-7. This test must be passed with a grade of 4,8 or higher to be used for the average. This test corresponds to 35% of the final grade of the subject.

• Multiple choice objective test of questions related to classroom practices. This test corresponds to 10% of the final grade of the subject.

2. Final exam: Students who are in the following situations may be presented to the final exam:

• Students who have obtained a grade below 4,8 of the theory part and seminars in any of the two partials.

• Students who have obtained a grade equal to or greater than 4,8 of the theory part and seminars in one or both partial tests but the average for this part of the evaluation does not reach 5.

• Students who want to upload a grade of one or both of the partials, or of the classroom practices or problems. The grade obtained in the final exam will be maintained.

This exam contains:

• Multiple choice objective test corresponding to each partial. The student will choose to perform one or both tests depending on their situation. This test must be passed with a grade of 4,8 or higher to be used for the average. Each test will correspond to 35% of the final grade of the subject.

• Objective written test of questionsrelated to classroom practices. This test corresponds to 10% of the final grade of the subject.

B. The competences acquired in the laboratory practices will be evaluated by continuous evaluation through a written test at the end of each practice. The average of the three tests corresponding to the three laboratory practices will be used to obtain the final grade. It is not necessary that the average of the three tests equal or exceed 5 to pass the course. Failure to show up to practice and, therefore, not perform the corresponding written test, represents a 0 in that laboratory practice.

The repeating students will only have to return to those lab sessions in which they have not reached a grade equal to or higher than 6 in the test of the corresponding practice, provided that this mark has been obtained in the last two years.

C. The final grade will be obtained as follows:

Theory and seminar tests: 0 70% of the final grade

Tests of classroom practices: 10% of the final grade

Tests of laboratory practices: 20% of the final grade

-Partial and final exams must reach marks of 4,8 or higher to be used for the average, as long as this average reaches 5.

-To pass the subject it will be necessary to obtain a global grade equal to or greater than 5 out of 10.

-The "Non-evaluable" will reflect the non-attendance to the final exam of recovery for students who have not passed the subject previously in the partial exams or who have to evaluate the whole subject through the final exam of recovery.

D. In the case that the student does not exceed the assessment requirements of the subject and its average grade of is greater than 5, the final grade cannot be higher than 4.8.

E. Those students with three failed calls (in the 3rd enrollment) may apply to perform a special synthesis exam that will include the entire subject,as long as they have been evaluated through the academic year..

Review of the exams

After each one of the exams of the subject, the review of the exam will be convened during which the students will be able to consult their exam and, if necessary, make a written and reasoned claim.

Single evaluation

Students undertaking this course may choose the single assessment system, according to the Faculty's regulations. The single assessment will be based on the same content course, the acquisition of the same skills, and will achieve the same level of demand as the continuous assessment.
The single assessment test will take place the same date fixed in the calendar for the last continuous assessment test and the same recovery system will be applied as for the continuous assessment. The single assessment will consist of the two tests corresponding to the two blocks of partials (including the classroom practice exam) and the tests corresponding to the three laboratory practices. The weight of each assessment activity will be the same as in the case of continuous assessment. To pass the subject through the single assessment system, each student must meet all the requirements indicated in the continuous assessment system.

Bibliography

Bibliography

Specific bibliography:

Peter D. Turnpenny, Sian Ellard. Emery's elements of medical genetics, 15th edition, Elsevier 2018, ISBN:9788491132066

Lynn B. Jorde, John C. Medical genetics. 5th ed., Elsevier, 2016, ISBN:9788491130581

Tom Strachan, Judith Goodship and Patrick Chinnery. Genetics and genomics in medicine. London : Garland Science, cop. 2015, ISBN:9780815344803

Tom Strachan and Andrew Read. Human molecular genetics. CRC Press, Taylor & Francis Group, 2019. ISBN:9780815345893

Ricki Lewis. Human genetics : concepts and applications. New York, NY : McGraw-Hill Education, 2018. ISBN:9781259700934

Robert L. Nussbaum, Roderick R. McInnes, Huntington F. Willard. Thompson & Thompson Genetics in Medicine. Elsevier 2016. ISBN:9781437706963

Bruce R. Korf, Mira B. Irons. Human genetics and genomics. 4th edition. Wiley-Blackwell, 2013. ISBN:9780470654477

T. A. Brown. Genomes 4. Garland Science, 2017, Fourth edition. ISBN:978081534508

Reference bibliography:

Lewis. Human Genetics. Concepts and applications. 9ª ed. McGraw-Hill International edition, 2010

Read A and Donnai D. New Clinical Genetics. 2nd edition. Scion Publishing Ltd, 2011

Internet resources:

http://www.nature.com/nature/supplements/collections/humangenome/index.html.

http://genome.wellcome.ac.uk/

http://www.ncbi.nlm.nih.gov/mapview/map_search.cgi?chr=hum_chr.inf&query

http://www.ncbi.nlm.nih.gov/genome/guide/human

http://www.ncbi.nlm.nih.gov/omim

http://www.geneclinics.org

Software

Miscrosoft programs, essentially PowerPoint, will be used to carry out the main lectures. Lecture presentation and PLAB booklet can be visualized with Adobe Reader.

CodonCode Aligner (Demo version): This software will be used on a PAUL.

The PLAB3 will use software previously installed on the computers in the computer room of the Faculty of Medicine (Campus Bellaterra).

If video conferencing is required, Teams will be used.

Language list

Name	Group	Language	Semester	Turn
(PAUL) Classroom practices	101	Catalan	first semester	afternoon
(PAUL) Classroom practices	102	Catalan	first semester	afternoon
(PAUL) Classroom practices	103	Catalan	first semester	afternoon
(PAUL) Classroom practices	104	Catalan	first semester	afternoon
(PAUL) Classroom practices	105	Catalan	first semester	afternoon
(PAUL) Classroom practices	106	Catalan	first semester	afternoon
(PAUL) Classroom practices	107	Catalan	first semester	afternoon
(PAUL) Classroom practices	108	Catalan	first semester	afternoon
(PAUL) Classroom practices	109	Catalan	first semester	afternoon
(PAUL) Classroom practices	110	Catalan	first semester	afternoon
(PLAB) Practical laboratories	101	Catalan	first semester	afternoon
(PLAB) Practical laboratories	102	Catalan	first semester	afternoon
(PLAB) Practical laboratories	103	Catalan	first semester	afternoon
(PLAB) Practical laboratories	104	Catalan	first semester	afternoon
(PLAB) Practical laboratories	105	Catalan	first semester	afternoon
(PLAB) Practical laboratories	106	Catalan	first semester	afternoon
(PLAB) Practical laboratories	107	Catalan	first semester	afternoon
(PLAB) Practical laboratories	108	Catalan	first semester	afternoon
(PLAB) Practical laboratories	109	Catalan	first semester	afternoon
(PLAB) Practical laboratories	110	Catalan	first semester	afternoon
(PLAB) Practical laboratories	111	Catalan	first semester	afternoon
(PLAB) Practical laboratories	112	Catalan	first semester	afternoon
(PLAB) Practical laboratories	113	Catalan	first semester	afternoon
(PLAB) Practical laboratories	114	Catalan	first semester	afternoon

(PLAB) Practical laboratories	115	Catalan	first semester	afternoon
(PLAB) Practical laboratories	116	Catalan	first semester	afternoon
(PLAB) Practical laboratories	117	Catalan	first semester	afternoon
(PLAB) Practical laboratories	118	Catalan	first semester	afternoon
(PLAB) Practical laboratories	119	Catalan	first semester	afternoon
(PLAB) Practical laboratories	120	Catalan	first semester	afternoon
(TE) Theory	101	Catalan	first semester	morning-mixed
(TE) Theory	102	Catalan	first semester	morning-mixed
(TE) Theory	103	Catalan	first semester	morning-mixed