

Degree programme	Type	Course
Biochemistry	OB	3

Contact lecturer

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Group languages

You can consult this information at the [end](#) of the document.

Prerequisites

There are no official prerequisites, but it is assumed that the student has previously acquired enough solid knowledge on subjects of 1st and 2nd degrees: Biochemistry I and II, Molecular Biology, Genetics and Basic and Advanced Instrumental Techniques.

Objectives

Provide a general knowledge about the molecular basis of the development of genetic diseases and deepen in the application of biochemical and molecular biology techniques for their study, diagnosis and therapeutics. In order to integrate this information, some selected examples of genetic diseases will be described at the molecular level.

Learning outcomes

- CM30 (Interpret clinical, histological, biochemical and molecular genetic data.) Interpret clinical, histological, biochemical and molecular genetic data.
- CM31 (Deliver a public presentation on a biomedical topic.) Deliver a public presentation on a biomedical topic.
- CM33 (Combine searches for new knowledge and problem-solving in biomedicine with ethical and social responsibility.) Combine searches for new knowledge and problem-solving in biomedicine with ethical and social responsibility.
- KM32 (Identify different types of animal tissue and cells.) Identify different types of animal tissue and cells.

- KM34 (Describe the molecular and pathophysiological foundations and biochemical markers of the most prevalent diseases in the population.) Describe the molecular and pathophysiological foundations and biochemical markers of the most prevalent diseases in the population.
- SM34 (Apply bioinformatics resources to analyse databases of genes and genetic diseases.) Apply bioinformatics resources to analyse databases of genes and genetic diseases.
- SM35 (Use histological, immunological and clinical laboratory techniques to study animal tissues and determine biochemical and genetic markers associated with different pathologies.) Use histological, immunological and clinical laboratory techniques to study animal tissues and determine biochemical and genetic markers associated with different pathologies.

Contents

THEORY

1. Introduction to genetic diseases. Definition of health and disease. Definition of genetic disease. Garrod's contribution: Inborn errors of metabolism. Data bases of genetic diseases. Monogenic, polygenic and multifactorial diseases. Mendelian inheritance. Incidence and prevalence of genetic diseases in the population.

2. Mutations in DNA as a cause of genetic diseases. Definition of mutation. Mutation rate. Types of molecular mutations and their effect on gene expression. Haemoglobinopathies. Enzymopathies: Blocking a metabolic pathway. Glucose-6-phosphatase, galactosemia and phenylketonuria deficiencies.

3. Molecular genetics diagnosis. Type and origin of analyzed samples. Prenatal and carrier diagnostics. Non-invasive techniques. Methods for detection of point mutations (SNPs), dynamic mutations, deletions and chromosomal rearrangements. Microarrays.

4. Molecular bases of inheritance and genetic diseases. Loss of function. Recessivity. Dominance. Haploinsufficiency. Negative dominant effect. Gain of function. Variable expressivity. Incomplete penetrance. Epigenetics. Genomic imprinting. Prader-Willi and Angleman syndromes. Chromosome X inactivation. Functional hemizygosity. Mosaic females. XIST gene and chromosome X inactivation center (XIC).

5. Identification of disease-associated genes. Strategies. Functional cloning. Positional cloning. Genetic and physical maps. Linkage analysis. LOD score. Zoo blots. CG isles. Exon trapping. Exon prediction. Chromosome jumping. Candidate genes.

6. Monogenic diseases: Cystic fibrosis. Alteration on chloride ion transport. Identification of the associated gene. Structure and function of the cystic fibrosis transmembrane regulator (CFTR). Effects of $\Delta F508$ and other mutations. Compound heterozygous. Therapeutic approaches.

7. Diseases caused by dynamic mutations. Classification. Proposed mechanism. General characteristics: Incomplete penetrance, anticipation, premutation. X-frangible syndrome. Effect of trinucleotide CGG expansion. Function of FMR1 gene.

8. Polygenic diseases: Alzheimer's disease. Types of lesions. Candidate and susceptibility genes. Amyloid

protein precursor (APP). Role of secretases in APP processing. Presenilins. Drugs: Acetylcholinesterase inhibitors. Additional therapeutic approaches.

9. Chromosomal diseases: Down syndrome (Trisomy 21). Effect of maternal age. Phenotype. Causes. Gene dosage effect. Candidate genes. Down syndrome critical region. Animal models. Prenatal diagnosis.

10. Diseases of amino acid metabolism. Phenylketonuria and other hyperphenylalaninurias. Phenylalanine hydroxylase deficiency. Structure and effect of mutations. Neonatal diagnosis and prevention.

11. Diseases of lipid metabolism. Familial hypercholesterolemia. Cholesterol metabolism and LDL. Associated loci. Structure and function of LDL receptor. Effect of mutations.

12. Diseases of carbohydrate metabolism. Glycogen storage diseases. Galactosemias.

13. Diabetes mellitus. Type 1 diabetes. Type 2 diabetes.

14. Lysosomal storage diseases. Enzyme replacement therapy.

15. Diseases of collagen biosynthesis and structure. Osteogenesis imperfecta. Ehlers-Danlos syndrome. Alport syndrome.

16. Muscular dystrophies. Duchenne muscular dystrophy. Becker muscular dystrophy. Limb-girdle muscular dystrophy. Structure of dystrophin and dystrophin-dystroglycan complex.

17. Biochemistry and molecular biology of cancer (I).

Cancer as a multicausal process. Hallmarks of cancer. Oncogenes. Relationship with signal transduction pathways.

18. Biochemistry and molecular biology of cancer (II).

Tumor suppressor genes: molecular basis and relationship with familial cancers. Relationship with DNA repair mechanisms.

19. Biochemistry and molecular biology of cancer (III).

Apoptosis, senescence and cancer. Angiogenesis. Tumor stroma. The immune system

20. Biochemistry and molecular biology of cancer (IV).

Molecular basis of invasion and metastasis.

21. Biochemistry and molecular biology of cancer (V).

Cancer therapy.

22. Biochemistry and molecular biology of cancer (VI).

Cancer therapy. Mechanisms of drug resistance. Risk factors.

23. Molecular biology techniques to study disease mechanisms (I). Introduction to techniques of transgenesis in animals. DNA microinjection into fertilized oocytes.

24. Molecular biology techniques to study disease mechanisms (II). Introduction to techniques of targeted genomic alterations: Obtaining knock-out and knock-in animals using classical and CRISPR / Cas9 techniques.

25. Introduction to gene therapy. Types of vectors. Development of strategies for gene transfer into specific cells and tissues.

SEMINARS

Suggested topics:

1. Pigmentary retinosis

2. Charcot-Marie-Tooth's disease

3. Wolfram Syndrome
4. Colorectal cancer
5. Lesch-Nyhan Syndrome
6. Amyotrophic lateral sclerosis
7. Adrenoleukodystrophy
8. Rett syndrome
9. Gaucher's disease
10. Malignant hyperthermia
11. Marfan Syndrome
12. Friedreich's Ataxia
13. Neurofibromatosis
14. Colorectal cancer
15. Mitochondrial diseases
16. Wolfram Syndrome
17. Narcolepsy
18. Schizophrenia

19. Alcoholism

20. Pharmacogenomics and toxicogenomics

PRACTICAL ACTIVITIES

The practical activities of this subject are carried out in the Integrated Laboratory 6 and consist of the following actions:

Genotyping, determination of cell viability and type of cell death and signaling.

- Extraction and purification of genomic DNA.
- Genotyping by PCR. Electrophoretic analysis.
- Determination of the lethal dose 50 of a drug on a human cell line.

Learning activities and methodology

Title	Hours	ECTS	Learning outcomes
Theory lectures	35	1.4	CM30, CM33, KM32, KM34, SM34, SM35
Delivery of clinical cases, short questions and problem solving, through the Intranet	12	0.48	CM30, CM33, SM34, SM35
Study	50	2	CM30, CM33, KM32, KM34, SM34, SM35
Tutorials	6	0.24	CM30, CM33, KM32, KM34, SM35
Preparation of seminars	26	1.04	CM30, CM31, KM34, SM34
Delivery of clinical cases, short questions and problem solving, through the Intranet	3	0.12	CM30, CM33, SM34, SM35
Seminars	10	0.4	CM30, CM31, KM34, SM34

Material available on the Intranet of the subject

Teaching guide

Calendar of teaching activities (lectures, tutorials, assessments, deliveries ...)

Visual material used by teachers in the lectures

Self-learning topics (Seminars)

Short questions, clinical cases, statements of problems

Collection test questions as an exam model

The training activities are divided into three sections: lectures and seminars, each one with its specific methodology. These activities will be complemented by a series of tutorial sessions that will be additionally scheduled and a collection of deliveries of tests for continuous evaluation.

Lectures

The content of different subjects will be explained with the support of visual material that will be available to students through the Intranet of the subject. This visual material will be written in Catalan, Spanish or English. Lecture sessions will be the most important part of the theory section.

Seminars

Knowledge of some parts chosen from the content of the different subjects will have to be searched through autonomous learning by students. It will be evaluated as oral presentations in the Seminar sessions and it will also be uploaded as a study material on the Intranet for all students to have access. Oral presentations done and presented in English.

The group will be divided into two subgroups (maximum 30 students per subgroup), whose lists will be made public at the beginning of the course. There will be 10 sessions of seminars during the course where students will prepare an oral presentation for a chosen self-study (see seminar contents). Presentations in PowerPoint format and a summary of a maximum half-page will have to be sent to the teacher a week before. She/He may suggest changes or modifications during that week that must be included in the presentation.

The oral presentation of the seminar will have a minimum duration of 20 min., with the following scheme:

- Inheritance and epidemiology,
- Clinical (symptomatology),
- Molecular genetics (chromosomal location and gene identification),
- Biochemistry (mutations / allelic variants and genotype-phenotype correlation),
- Diagnosis and therapeutics.

The rest of the time will be devoted to solving doubts, answering questions, raising a debate, etc., where all those attending the seminar will be able to participate.

The oral presentation will be shared among the members of the group (2 students), so that everyone has the opportunity to speak at least for 10 min.

Attendance and accomplishing oral presentations at the Seminars are mandatory for all students, except in cases where there is a documented justification. Active participation of the students in the seminars will be rated, so that it will have an impact on the seminar grade. Lack of attendance will discount a percentage of the seminar mark.

Delivery of clinical cases, short questions or problems

Every 10-12 theory topics a collection of questions that may contain clinical cases, short questions or problems will be delivered through the intranet tool and that will have to be answered back within a short period of time that will be defined at the proposal. Questions will be related to concepts explained in the lectures but also to self-learning topics that must be searched and studied through autonomous learning by the students.

Tutorials

Tutorials will be carried out as requested by students. If the number of requirements is extremely high, especially for midterm exams or the resolution of clinical cases or short questions, up to 3 classroom tutorials would be scheduled and they will be announced on a timely basis through the intranet. The objective of these sessions will be to resolve doubts, review basic concepts, solve problems or clinical cases proposed through intranet short questions, guide on the consulted sources of information and carry out debates on the topics for which there are planned autonomous learning or that have been proposed by the teachers. These sessions will not be lectures nor will be treated new topics from the official content of subjects, but will be sessions of debate and discussion.

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

Continuous assessment activities

Title	Weight	Hours	ECTS	Learning outcomes
Delivery of clinical cases, short questions and problem solving, through the Intranet	10%	1	0.04	CM30, CM31, KM34, SM34
Theory midterm individual exams	65%	6	0.24	CM30, CM33, KM32, KM34, SM34, SM35
Oral presentation and participation in seminars	25%	1	0.04	CM30, CM33, SM34, SM35

Continuous Evaluation

The evaluation of this course will have the format of continuous assessment with a re-assessment final test. The objective of the continuous assessment is to encourage the students' effort throughout the semester, allowing them to monitor their degree of follow-up and understanding of the subject. The final test of re-assessment is used to verify that the student has reached the necessary degree of integration of knowledge of the course. To be eligible for the retake process, the student should have been previously evaluated in a set of activities equaling at least two thirds of the final score of the course or module. Thus, the student will be graded as "No Avaluable" if the weighthin of all conducted evaluation activities is less than 67% of the final score.

Any irregularity committed during an assessment activity (academic misconduct, plagiarism, or improper use of AI, unless such use is expressly authorized in the course syllabus) that may lead to a significant alteration of the grade will result in that activity being graded as 0. If the course syllabus stipulates that obtaining a minimum mark in this assessment is an essential requirement to pass the course, or if multiple irregularities occur in the assessment activities of the same course, the final grade for the course will be 0. Furthermore, disciplinary proceedings may be initiated against any student who incurs any of these irregularities.

Seminars

Assessment of teamwork:

The obtained mark will be the same for all the members of the team, as long as all of them have prepared and exhibited in an equivalent manner. The involvement of the different members of the team will be verified through a small individual and confidential survey. The weight of the evaluation of teamwork will be 25% of the total. In order for students to acquire transversal skills, the seminar must be taught in English; the use of another language will be penalized with 2.5 points.

Evaluation of individual learning:

The two sections (Theory and Seminars) are inseparable, so that the student must participate, and be evaluated, in both to overcome the matter. Therefore, participation in the seminars is mandatory, both on the day of the oral presentation and the attendance at the other seminars of the peers. Students missing more than 50% of programmed sessions will be graded as "No Avaluable". The active participation of the students in the seminars will have an impact on the seminar grade. A selection of seminars, which are also part of the contents of the subject, will be evaluated with a question to the examination of the 2nd midterm exam, and will contribute in a proportional way to the mark of this exam. The re-assessment exam will also include a seminar question.

Theory

Individual assessment through:

- Two midterm tests with short questions.
- A final proof of re-assessment that will have the same format as the midterm exams and will cover the entire subject of the course. This exam is intended for students who have not previously passed midterm tests.
- Delivery of answers for clinical cases, problems and continuous assessment tests through the intranet (maximum 2 deliveries)

The weight of the theory evaluation will be 75% of the total. Of this, 10% will correspond to the score of the delivery of answers through the intranet, and 65% to the theory exams (32.5% for each midterm exam).

In order to be able to do the average between midterm tests, without going to the final re-assessment test, the student will have to obtain in the two midterm exams a qualification equal or higher than 4.5. The topics corresponding to the partial theory tests with a qualification of less than 4.5 will be evaluated in the final re-assessment test, where it will be also necessary to obtain a qualification equal or higher than 4.5 of each midterm topics to be able to average with the scores of the rest of the activities. However, those students who have passed the partial tests of theory and want to improve their qualification may choose to attend to the final test of re-assessment for the totality of the subject or only one of the midterm exams. The student who is attending a re-assessment exam is resigning the partial / s scores.

It is necessary to obtain a final grade equal to or greater than 5 to pass the course, either through midterm or through the final re-assessment test.

It will be considered that a student will obtain the "not evaluated" qualification when the number of assessment activities carried out is less than 67% of those programmed for the course.

The students from a second enrolment of the course will not have to carry out the educational activities or the evaluations of those activities passed with a score higher than 5, like the seminars and the delivery of questions of continuous assessment.

Unique Evaluation

Students taking the unique assessment must present their seminar (SEM) on the day that corresponds to them according to the seminar calendar that will be announced through the Virtual Campus. The assessment and weight on the final grade of this activity will be the same as those of the continuous assessment (SEM 25%).

The single assessment consists of a single summary test (with short questions) on the contents of the entire theory program. Both, in the synthesis test as in the re-assessment exam there will also be a seminar question.

The mark obtained in the synthesis test and in the re-assessment test is 65% of the final mark of the subject, the mark obtained in the questionnaires delivered through the Virtual Campus is 10% and the seminars, the remaining 25%.

The single assessment test will coincide with 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.

To pass the subject you must obtain a final grade of at least 5 points out of 10 in the overall grade of the subject and to be able to average the marks of the different activities a minimum of 4.5 must be obtained in the synthesis and re-assessment tests.

It will be considered that a student will obtain the "not evaluated \" qualification when the number of assessment activities carried out is less than 67% of those programmed for the course.

The students from a second enrolment of the course will not have to carry out the educational activities or the evaluations of those activities passed with a score higher than 5, like the seminars and the delivery of questions of continuous assessment.

Bibliography

Basic Bibliography

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Complementary Bibliography

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Internet addresses

On-line Mendelian Inheritance in Man (OMIM). <http://www.ncbi.nlm.nih.gov/Omim/>

Molecular Medicine MedPulse®. <http://www.medscape.com/px/splash>

Medline Plus®. <http://www.nlm.nih.gov/medlineplus/>

Genes and Disease.

<http://www.ncbi.nlm.nih.gov/books/bv.fcgi?call=bv.View..ShowTOC&rid=gnd.TOC&depth=2>

Software

None

Course groups and languages

The information provided is provisional until November 30. After this date, you will be able to consult the language of each group through this [link](#). To access the information, you will need to enter the course CODE

Type of teaching	Group	Language	Semester	Shift
(TE) Theory	33	Catalan	second semester	morning-mixed
(PAUL) Classroom practices	331	English	second semester	morning-mixed
(PAUL) Classroom practices	332	English	second semester	morning-mixed