

Academic Year/course: 2021/22

## 27116 - Clinical Biotechnology

### Syllabus Information

**Academic Year:** 2021/22

**Subject:** 27116 - Biotecnología clínica

**Faculty / School:** 100 - Facultad de Ciencias

**Degree:** 446 - Degree in Biotechnology

**ECTS:** 9.0

**Year:** 3

**Semester:** Annual

**Subject Type:** Compulsory

**Module:**

## 1. General information

### 1.1. Aims of the course

Main aims:

To cover the biotechnology applications in the clinical field, as well as, to emphasize the role of biotechnology in accelerating the understanding of the molecular basis of disease. This course deepens into the physiopathology, diagnosis, therapeutic approaches and treatment of diseases.

## 2. Learning goals

## 3. Assessment (1st and 2nd call)

## 4. Methodology, learning tasks, syllabus and resources

### 4.1. Methodological overview

**The learning project of this subject is based on:**

A reinforcement of theoretical bases with involvement of students through a guided supervision. Students will prepare a subject of their interest and related to topics covered in this subject. Special emphasis will be paid to research aspects with new and innovative approaches.

### 4.2. Learning tasks

The course includes the following learning tasks:

- Lectures. Presential. 6 ECTS. Provide students basic background of the subject. Presentations, animations, videos and surfing the net will be elements used. Didactic materials will be available for the students through the private server of UNIZAR
- Experimental work. 2 ECTS. This activity will provide training in laboratory analysis and data management.
- Seminars. Presential. 0.5 ECTS. Case-oriented approaches will deepen basic concepts of the subject
- Presentation and discussion of a selected case. 0.5 ECTS. Students will review a particular topic in close supervision by a faculty member. This guidance will be set by appointed meetings. Eventually and in an open

session, students will deliver their presentations to their mates and faculty members.

### 4.3. Syllabus

The course will address the following topics:

#### Lectures

1. Introduction to Clinical Biotechnology. Semiology. Definition of syndrome. Overview of clinical syndromes.
2. Diseases of carbohydrate metabolism. Classification of carbohydrate metabolism disorders. Deficiencies of intestinal glycosidases.
3. Pentosurias. Primary hyperoxalurias.
4. Disorders of galactose metabolism. Galactosemias. Hereditary fructose intolerance. Fructose metabolism disorders
5. Glucose metabolism disorders: Diabetes mellitus type I and II. Sequelae of diabetes mellitus.
6. Glycogen metabolism disorders. Glycogenesis.
7. Metabolism of lipoproteins. Overview of lipoprotein metabolism. Primary and secondary dyslipidemias. Classification.
8. Disorders affecting lipoprotein metabolism: Hyperchilomicronemias. Type III Hyperlipoproteinemia. Familial combined hyperlipidemia.
9. Hypercholesterolemias. Autosomal dominant hypercholesterolemia. Hypoalphalipoproteinemias.
10. Disorders of protein metabolism. Overview of amino acid metabolism. Amino acid transport disorders.
11. Disorders of ammonium metabolism. Enzyme deficiencies in the urea cycle.
12. Aminoacidopathies: Alcaptonuria. Albinism. Phenylketonuria. Tyrosinosis. Maple syrup urine disease. Other aminoacidopathies.
13. Disorders of the metabolism of purines and pyrimidines. Uric acid. Primary and secondary hyperuricemia.
14. Lysosomal storage disorders (I): Mucopolidosis and mucopolysaccharidosis.
15. Lysosomal storage disorders (II). Sphingolipidoses and other lysosomal diseases.
16. Landscape of research in Clinical Biotechnology.
17. Molecular bases of mitochondrial diseases.
18. Peroxisomal diseases.
19. Pathologies associated with oxidative stress. Molecular bases and therapeutic strategies.
20. The nucleic acids as therapeutic agents. Background. Oligonucleotidos and antisense RNA. Ribozymes. Aptamers. Current status and approaches.
21. Therapy using siRNA. Introduction. Background. Requirements. Methods. siRNA as antiviral therapy. HIV. siRNA as antitumor therapy. siRNA in other diseases. Prospects.
22. Disorders in iron metabolism and hemoglobin. Anemia and poliglobulias. Hemochromatosis. Hemoglobinopathies. Thalassemia. Porphyrin metabolism. Porphyrria.
23. Hemostasis disorders. Hypercoagulability status and hemorrhagic diathesis.
24. Laboratory tests of kidney function (cystatin C, creatinine clearance, PAH, osmolarity, free water etc ...) and urinalysis. Renal pathophysiology: failure, and nephrosis.
25. Biochemical markers of ischemic heart disease. Heart failure biomarkers.
26. Liver pathophysiology. Methods for assessing liver function. Analysis of the bile duct integrity.
27. Laboratory tests of gastric, pancreatic and intestinal function. Gastric and exocrine pancreatic pathophysiology.
28. Bone metabolism pathophysiology: calcium, magnesium and phosphate. Hypercalcemia, hypocalcemia, hyperparathyroidism and hypoparathyroidism.

29. Analytical exploration of pituitary hormones. Pathophysiology of gigantism, acromegaly and dwarfism. Basal and dynamic tests. Pathophysiology of prolactin disorders.
30. Analytical exploration of the neurohypophyseal hormones. Pathophysiology of diabetes insipidus.
31. Analytical exploration of the hypothalamic-pituitary-adrenal gland axis. Analytical examination of the adrenal glands.
32. Pathophysiology of Cushing's syndrome and Addison disease.
33. Analytical exploration of thyroid hormones. Pathophysiology of hyperthyroidism and hypothyroidism.
34. Pathophysiology of hypothalamic pituitary axis. Sex glands. Analytical study of male and female infertility.

#### Seminars

- 1. Autosomal dominant hypercholesterolemias
- 2. Anderson-Fabry disease.
- 3. Treatment of mitochondrial diseases.
- 4. Cornelia de Lange syndrome. Genetic bases.

#### Practice sessions

- 1. Dyslipidemia: APOE genotyping
- 2. Biomarkers in lysosomal disorders. Plasma chitotriosidase activity. CHIT genotype
- 3. Genetic diagnosis of Gaucher disease.
- 4. Laboratory of Clinical Biochemistry. Quality control.
- 5. Plasma follow-up of hepatic function
- 6. Urinalysis.

#### **4.4. Course planning and calendar**

Scheduled sessions and exam dates will be available through the Web of Facultad de Ciencias, Biotechnology section, at the following url: <https://ciencias.unizar.es/grado-en-biotecnologia>.

At the beginning of the academic year, the Degree' coordinator will set different groups of experimental sessions and their schedule in order to avoid overlapping with other subjects.

#### **4.5. Bibliography and recommended resources**

<http://psfunizar10.unizar.es/br13/egAsignaturas.php?codigo=27116>