



Universitat de Lleida

DEGREE CURRICULUM  
**MOLECULAR PATHOLOGY AND  
BIOMEDICAL DIAGNOSTIC**

Coordination: TORRES ROSELL, JORDI

Academic year 2023-24

# MOLECULAR PATHOLOGY AND BIOMEDICAL DIAGNOSTIC 2023-24

## Subject's general information

<b>Subject name</b>	MOLECULAR PATHOLOGY AND BIOMEDICAL DIAGNOSTIC			
<b>Code</b>	101626			
<b>Semester</b>	2nd Q(SEMESTER) CONTINUED EVALUATION			
<b>Typology</b>	<b>Degree</b>	<b>Course</b>	<b>Character</b>	<b>Modality</b>
	Bachelor's Degree in Biotechnology	4	OPTIONAL	Attendance-based
	Master's Degree in Biomedical Research		COMPLEMENTARY TRAINING	Attendance-based
<b>Course number of credits (ECTS)</b>	6			
<b>Type of activity, credits, and groups</b>	<b>Activity type</b>	PRALAB	PRAULA	TEORIA
	<b>Number of credits</b>	1	0.8	4.2
	<b>Number of groups</b>	1	1	1
<b>Coordination</b>	TORRES ROSELL, JORDI			
<b>Department</b>	BASIC MEDICAL SCIENCES			
<b>Teaching load distribution between lectures and independent student work</b>	60 hores presencials 90 hores no presencials			
<b>Important information on data processing</b>	Consult <a href="#">this link</a> for more information.			
<b>Language</b>	Català Anglès			

Teaching staff	E-mail addresses	Credits taught by teacher	Office and hour of attention
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## Subject's extra information

Aquesta assignatura es situa com a optativa de quart curs de Biotecnologia per tal de donar a conèixer les metodologies analítiques emprades actualment en els laboratoris d'anàlisi clínic i genètic, així com els fonaments bàsics de les alteracions metabòliques i les malalties associades a aquestes alteracions. L'assignatura també introdueix els fonaments de les patologies d'origen genètic i les eines actuals emprades en el seu diagnòstic, fent èmfasi en el càncer com a exemple de patologia genètica. Amb aquesta finalitat es descriuen els mecanismes de regulació del cicle cel·lular i l'estabilitat genòmica, i s'aprofundeix en l'anàlisi de la patologia del càncer des del punt de vista molecular.

## Learning objectives

1. Acquire the basic knowledge on the background and interpretation of laboratory tests in the field of Clinical Biochemistry.
2. The student will understand the relationship between the molecular mechanisms of control and the pathology that derives from genetic alteration.
3. Understand and know the main principles of genetic diagnosis.
4. As a paradigmatic example of molecular pathology, understand the fundamental discoveries that have helped us to understand the molecular biology of cancer.
5. In a more applied way, understand the development and progression of cancer from the point of view of

## Competences

- GC1 Be able to selectively search for and use sources of information necessary to achieve the training objectives.
- GC2 Interpret scientific-technical information with a critical sense, and be able to make presentations based on this information.
- GC4 Knowing and adequately using the scientific and technical vocabulary of the different areas of Biotechnology.
- GC5 Working in the laboratory applying criteria of quality and good practice.
- GC11 Acquiring criteria for choosing the most appropriate analytical techniques for each specific practical case.

## Subject contents

### MODULE 1. CLINICAL BIOCHEMISTRY.

Topic 1: Glucose homeostasis. Study of diabetes mellitus. Methodology for the determination of glucose in biological fluids. Glucose tolerance test. Clinical importance of ketone bodies.

Topic 2: Anomalies in intestinal carbohydrate metabolism: Intolerances due to deficiencies in disaccharidases.

Topic 3: Plasma proteins: Albumin, Immunoglobulins and other proteins of clinical interest. Proteinograms. Protein in urine.

Item 4-5: Metabolism of nitrogen compounds: Amino acids and nucleotides. Diagnostic value of transaminases. Urea cycle defects and related disorders causing hyperammonemia. Creatine and creatinine. Alterations in purine base metabolism: hyperuricemia.

Item 6-7: Composition and metabolism of plasma lipoproteins. Disorders of Lipoprotein Metabolism: Relationship with Atherosclerosis. Cholesterol levels and cardiovascular risk factors. Control of dyslipidemias. Analytical methods.

Topic 8: Heme group metabolism. Diseases related to the metabolism of the heme group. Porphyria: types and consequences. Bilirubin: training, determination and diagnostic value.

### MODULE 2. GENETIC DIAGNOSIS.

Item 9: Genetic diagnosis. (2h). Pre-implantation diagnosis. Postnatal diagnosis. Genetic diagnosis of chromosomal disorders. Chromosomal identification techniques. Karyotype. Cytogenetics. FISH. Genomic hybridization.

Topic 10: Case study of genetic diagnosis based on mass sequencing techniques (NGS) (8h)

### MODULE 3. CELL PROLIFERATION AND GENOME STABILITY.

Topic 11: Introduction: from molecular bases to pathology. Beginnings of Molecular Medicine and Molecular Pathology. Classification of molecular pathologies. Examples of molecular pathologies.

Topic 12: Introduction to the cell cycle. Mechanisms and strategies for controlling cell division.

Item 13: Elements and mechanisms involved in chromosome replication and segregation.

Item 14: Control of cell proliferation and growth.

Topic 15: Molecular bases of pathologies that affect DNA repair. Checkpoints. Response to DNA damage.

Item 16: Genomic instability.

Item 17: Cell cycle and cancer as an example of genetic pathology.

### MODULE 4. PATHOLOGICAL ANATOMY OF CANCER.

Item 18: General principles of cancer. Terminology. Definitions. Benignity and Malignancy.

Topic 19: Natural History of Cancer. Invasion and Metastasis.

Topic 20: General Concepts About the Pathological Anatomy of Cancer. Epithelial tumor. Mesenchymal, nervous and melanoma tumors.

Item 21: Hematological tumors.

Item 22: Morphological diagnosis of cancer. Biopsies. Cytology. 2h.

Item 23: Immunohistochemical techniques in the diagnosis of cancer.

Topic 24: Molecular basis of cancer. Oncogenes, tumor suppressor genes, DNA repair genes (I). 2h.

Item 25: Molecular diagnosis of sporadic cancer. Somatic alterations 2h.

Item 26: Molecular diagnosis of familial cancer. Germ alterations.

Item 27: Immunohistochemical and molecular techniques in cancer prognosis.

Item 28: Molecular techniques in predicting the response to antineoplastic treatment. Therapeutic targets.

## PRACTICE MODULE 1

2 practical sessions.

1) Determination of total cholesterol, HDL and serum triacylglycerides

2) Determination of total iron in serum

## PRACTICE MODULE 2 (2h)

Computer class session

Practical exercise: Design of a diagnostic strategy using online resources.

## SEMINARS MODULE 3 (4h)

Study of six classical molecular pathologies: Clinical manifestations, isolation of the responsible gene, function of the wild-type gene, described mutations, pathological alterations of gene function and therapies. As a guide, the following will be studied:

1) Cornelia de Lange Syndrome

2) Fanconi's anemia

3) Bloom syndrome

4) Xeroderma pigmentosum

5) Telangiectasia ataxia

6) Family Breast Cancer (Genes BRCA1 and BRCA2)

## SEMINAR MODULE 4 (2h)

Students will be provided with a powerpoint to work with information on "Cancer Research Techniques."

## Methodology

Most of the subject is developed in 2-hour theoretical concept exposition sessions.

Some part of the agenda may be completed with videos and discussion sessions.

These concepts are exemplified in practical sessions and reinforced in seminar sessions with oral presentation. To achieve the objectives and acquire the assigned skills, the following activities will be scheduled:

Clinical biochemistry 14h

Cell cycle and genome stability 12h

Genetic diagnosis 8h

Pathological Anatomy of Cancer 14h

## LABORATORIES

Clinical Biochemistry Internship 4h

## SEMINARS

Pathologies of molecular origin seminars 4h

Pathological anatomy seminars 2h

## COMPUTER LABORATORY

Design of a diagnostic strategy 2h

Due to the COVID pandemic situation, some activities such as keynote sessions may be virtualized.

## Development plan

Acquire basic knowledge on fundamentals and interpretation of laboratory tests in the field of Clinical Biochemistry  
7 theory classes of 2h (single group)

Know the fundamental findings that have helped to understand the molecular biology of cancer. 6 theory classes of 2h (single group)

Know the main tools for genetic diagnosis of pathologies 4 theory classes of 2h (single group)

Understand how cancer develops and progresses from the point of view of pathological anatomy. 7 theory classes of 2h (single group)

Practices of Clinical Biochemistry 2 sessions of 2h

Analysis, study and presentation of the molecular bases of six pathologies 2 sessions of 2h

Practices in the computer room: Design of a diagnostic strategy 1 session of 2h

Know and understand cancer diagnostic methods 1 session of 2h

## Evaluation

There will be two test-type exams, one at the end of the first module, the second on the last day of class.

In addition, Module 2 will be evaluated through a paper that must be submitted well in advance to the teacher in charge.

Module 1 (30%) test type

Module 2 (17%) Written work

Modules 3 and 4 (46%) test type

Seminars Module 3 (7%) Presentation in the classroom

The evaluation system may vary depending on the health situation, being face-to-face, non-face-to-face (online) or a combination of both.

## Bibliography

Principios de Bioquímica Clínica y Patología Molecular. A. Gonzalez-Hernandez. 2010. Elsevier.

Tietz textbook of Clinical Chemistry. C.A. Burtis and A.R. Ashwood. Saunders. 3rd.edition.

González-Sastre i Joan J. Guinovart, (2000). *Lliçons de Patologia Molecular*. Springer.

Alberts B, et al. (2007), *Molecular Biology of the Cell*. Garland Science

Lewin B (2007), *Genes IX*. Jones & Bartlett

Morgan D. (2007). *The Cell Cycle: Principles of Control*. New Science Press Bronchud MH, (2008) *Principles of Molecular Oncology*. Humana Press ROBBINS y COTRAN (2005). *Patologia Estructural y Funcional*. Elsevier