

**COURSE DATA****DATA SUBJECT**

Code: 34496
Name: Molecular principles of pathology
Cycle: Undergraduate Studies
ECTS Credits: 4.5
Academic year: 2025-26

STUDY (S)

Degree	Center	Acad. year	Period
1204 - Degree in Medicine	Facultat de Medicina i Odontologia	4	First quarter

SUBJECT-MATTER

Degree	Subject-matter	Character
1204 - Degree in Medicine	Optional subjects	ELECTIVES

COORDINATION

DONATO MARTIN MARIA TERESA

SUMMARY

The objective of this subject is to provide students with knowledge on Molecular Pathology and Clinical Biochemistry which doctors should acquire. On the one hand, students will study genes which codify proteins the alteration of which produces diseases, and they will analyse the structure and the function of such proteins, trying to establish genotype-phenotype links which may explain why certain pathologies appear. On the other hand, biochemical exams will be seen as support tools for the diagnosis of diseases, facilitating the identification and the characterisation of pathological processes.

PREVIOUS KNOWLEDGE**RELATIONSHIP TO OTHER SUBJECTS OF THE SAME DEGREE**

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

OTHER REQUIREMENTS

It is recommended that students have passed the first 2 academic years of the Degree in Medicine.



COMPETENCES / LEARNING OUTCOMES

-

Acknowledge diversity and multiculturality.

Capacity for communicating with professional circles from other domains.

Consideration of ethics as a fundamental value in the professional practise.

Criticism and self-criticism skills.

Proper organisation and planning of the workload and timing in professional activities.

Students must be able to apply their knowledge to their work or vocation in a professional manner and have acquired the competences required for the preparation and defence of arguments and for problem solving in their field of study.

Students must have the ability to gather and interpret relevant data (usually in their field of study) to make judgements that take relevant social, scientific or ethical issues into consideration.

Team-working skills and engaging with other people in the same line of work or different.

Understand and recognise the structure and normal function of the human body, at the following levels: molecular, tissue, organic, and of systems, in each phase of human life and in both sexes.

Working capacity to function in an international context.

DESCRIPTION OF CONTENTS

THEORETICAL LESSONS.

1. The molecular bases of the disease. Genetic diseases.

Concept of Clinical Biochemistry. Molecular Pathology as an area of knowledge. Molecular level in medical pathology. Metabolopathy concept. Types of genetic alterations. Diagnostic strategies for genetic diseases.

2. Biochemical and molecular basis of metabolic disorders in carbohydrate and its diagnosis.

The role of the laboratory in the differential diagnosis of glucidic disorders. Inherited diseases of the carbohydrate metabolism. Diabetes; glycogen storage disease, carbohydrate intolerance.



3. Biochemical and molecular basis of lipidic disorders and their analytical techniques.

Blood markers of dyslipidaemias. Molecular disorders of enzymes, apolipoproteins, and membrane receptors. Molecules involved in fat storage and in the molecular pathology of obesity. Molecular diagnosis of hypercholesterolemia.

4. Clinical enzymology.

The role of enzymes in clinical diagnosis. Measurement of enzymes in serum. Enzymes of clinical interest. Enzymatic profiles. Isoenzymes and their diagnostic value.

5. Plasma proteins.

Plasma proteins: properties and functions. Methods of protein screening in plasma. Electrophoretic standards. Diagnostic implications. Studies on specific proteins.

6. Diagnosis of the liver diseases in the laboratory.

Hepatic functions and their role in the homeostasis of the organism. Liver Function Test. Biochemical profiles of acute hepatitis (viral, toxic), chronic, cirrhosis. Alcoholic Liver Disease. Intrahepatic and extrahepatic cholestasis. Hyperbilirubinemias.

7. Diagnosis of Kidney/ urine disease.

Biochemical integration of the Kidney. The renal functions. Nephrotic and nephritic syndrome. Renal failure and its evaluation by the laboratory. Physical and physicochemical characteristics of urine. Emergence of abnormal metabolites and its relationship with certain illnesses. Screening for proteinuria. Urinary sediment test. Biochemical assessment of the renal functional.

8. Diagnosis of cardiovascular diseases.

Biochemistry of the heart muscle. Epidemiology and Etiopatogenicity of cardiovascular diseases. Biochemical markers of interest and their interpretation.



9. Tumour markers.

Biochemical and molecular characteristic of tumour cells. Cell transformation markers: oncogenes and tumour suppressor genes. Protein tumour markers.

10. PRACTICAL LESSONS

CLASSROOM SEMINARS PROGRAM

1. Omics (Genomics, Transcriptomics, Proteomics, and Metabolomics): basis and their application in clinical diagnosis.
2. Aminoacidopathies. Concept and classification. Alterations in the metabolism of amino acids due to enzyme or transport protein defects. Diagnosis and treatment. Hyperphenylalanemia. Cystinuria.
3. Alterations in the metabolism of nitrogenous bases. Purine metabolism. Congenital disorders of purine metabolism. Mechanistic study of hyperuricemia.
4. Biochemical determinations in extravascular biological fluids. Analysis of serous fluids, cerebrospinal fluid, synovial fluid, etc. and their diagnostic applications.
5. Biochemical and molecular bases of clinical toxicity. Metabolism, detoxification and bioactivation of xenobiotics. Mechanisms of toxicity. Molecular targets. Cellular targets. Final consequences.
6. Biological specimens; collection and their analysis. The laboratory in the context of clinical diagnosis. Phases of analytical process in the clinical laboratory.
7. The organization of the analytical laboratory in a hospital. Quality controls, errors, and analytical interferences. Laboratory safety.

CLINICAL PRACTICES

Practice in hospital clinical diagnostic laboratories. Stay in a routine and/or emergency clinical biochemistry laboratory, and follow-up of the comprehensive analytical process. 4 hours.

WORKLOAD

PRESENCIAL ACTIVITIES

Activity	Hours
Theory	19,00
Seminars	18,00
Laboratory	0,00
In-class tutorials	4,00
Clinical practice	4,00
Total hours	45,00

**NON PRESENCIAL ACTIVITIES**

Activity	Hours
Attendance at other activities	0,00
Individual or group project	24,00
Independent study and work	37,50
Preparation of lessons	0,00
Preparation for assessment activities	6,00
Resolution of case studies	0,00
Preparation of supplementary reports	0,00
Preparation of the internship report and evaluation of the internship	0,00
Total hours	67,50

TEACHING METHODOLOGY

In the **theoretical lessons**, the teachers will present the most important concepts and contents of the subject in a structured manner, to promote the acquisition of knowledge and skills by the students. Student participation in these sessions will be encouraged. The teaching materials used by the professor will be made available, if deemed appropriate, through the electronic resource of the Virtual Classroom.

Classroom Practices: Seminars. In small groups, the professor will delve into specialized topics in depth and case studies or specific examples. Group work and cooperative learning will be encouraged.

Clinical Practices: Stay of the students in the clinical diagnostic laboratory service of university hospitals supervised by a professor. Students will learn how a routine and/or emergency clinical biochemistry laboratory works and how to monitor the different stages of the analytical process.

Scheduled Tutorials. Preparation and oral discussion by the students, individually or in small groups, of clinical cases or a work related to the subject proposed and supervised by a professor in charge. Tutorial sessions will also serve to resolve any doubts that may have arisen throughout the various learning activities and to monitor the learning process.

The gender perspective, the respect for diversity, and the sustainable development goals (SDGs) will be incorporated into teaching, whenever possible.

EVALUATION



A written test will be conducted with the aim of jointly assessing the acquisition of knowledge from the theory program, seminars, and clinical practices. The written test will consist of short-answer questions (50% of the exam grade) and multiple-choice questions (50% of the exam grade) with 4 answer options; in the latter, each incorrect response will incur a penalty of 25% of a correct answer, while blank responses will not be penalized. This written test will account for 85% of the subject's grade.

Evaluation of scheduled tutorials (15% of the final grade): Continuous assessment of participation and work carried out in these groups.

Attendance at clinical laboratory practice is mandatory to pass the subject.

The subject is passed with an overall grade of 5. It is not necessary to pass each part separately (theory, practices, tutorials).

Attendance at practical activities is mandatory. The student is considered to meet this requirement if he or she has attended a minimum of 80% of these activities and has adequately justified the impossibility of attending the remaining sessions due to the occurrence of a cause of force majeure. It will be essential to comply with this requirement to pass the subject.

Students are reminded of the importance of carrying out evaluation surveys on all the teaching staff of the degree subjects.

REFERENCES

- Marshall, W. J.: Bioquímica clínica, 7^o edición. Elsevier España, 2013.
- Murphy, M.: Bioquímica clínica. Texto y atlas en color., 6^o edición, Elsevier España, 2020.
- González Hernández, Á.: Principios de bioquímica clínica y patología molecular, 3^a edición, Elsevier España, 2019.
- González de Buitrago, J.M. et al.: Patología molecular. McGraw-Hill Interamericana, 2001.
- Fuentes Arderiu, X. et al.: Bioquímica clínica y patología molecular. Ed. Reverté, 1998.
- Scriver, C.R. et al.: The metabolic and molecular bases of inherited diseases. McGraw-Hill Book, 2001.
- Burtis, C.A. et al.: Tietz fundamentals of clinical chemistry. Ed. Saunders Elsevier, 2001.
- Herrera E. Bioquímica básica ISBN 10: 8480868988 / ISBN 13: 9788480868983. Editorial: Elsevier Editorial, 2014.
- Gaw, A.: Bioquímica clínica: texto ilustrado en color. Elsevier, 2001.
- RECURSOS e-Salut: ClinicalKey Student Medicina, Odontología y Enfermería [<https://uv-es.libguides.com/RecursosSalut>] Acces Medicina [https://uv-es.libguides.com/Access_Medicina] Médica Panamericana [https://uv-es.libguides.com/Medica_Panamericana]
- Baynes, J.W.: Bioquímica médica. 5^o edición Elsevier, 2019.