

**COURSE DATA****DATA SUBJECT****Code:** 33961**Name:** Molecular Pathology**Cycle:** Undergraduate Studies**ECTS Credits:** 4.5**Academic year:** 2025-26**STUDY (S)**

Degree	Center	Acad. year	Period
1201 - Degree in Pharmacy	Facultat de Farmàcia i Ciències de L'alimentació	5	Sin determinar, Second quarter
1205 - Degree in Human Nutrition and Dietetics	Facultat de Farmàcia i Ciències de L'alimentació	2	Sin determinar, Second quarter
1211 - Double Degree in Pharmacy and Human Nutrition and Dietetics	Facultat de Farmàcia i Ciències de L'alimentació	5	Sin determinar, Second quarter

SUBJECT-MATTER

Degree	Subject-matter	Character
1201 - Degree in Pharmacy	Molecular Pathology	ELECTIVES
1205 - Degree in Human Nutrition and Dietetics	Molecular pathology and physiopathology	COMPULSORY
1211 - Double Degree in Pharmacy and Human Nutrition and Dietetics	Asignaturas obligatorias del PDG Farmacia-Nutrición Humana y Dietética	COMPULSORY

COORDINATION

GUASCH AGUILAR ROSA

SUMMARY

Molecular Pathology is a compulsory second year course (second semester) in the degree of Human Nutrition and Dietetics and in the fifth year (second semester) in the degree of Pharmacy and Nutrition and Dietetics, which is taught in the Faculty of Pharmacy, University of Valencia. This course provides the curriculum for a total of 4.5 ECTS.

The main objective of the course is to understand the molecular and biochemical basis of major endocrine-metabolic syndromes, as well as some of the organic-functional disorders more common in the human population.

The minimum concepts acquired by the student of Molecular Pathology include:



- Basic concepts in molecular pathology.
- The molecular basis of disease.
- Techniques of molecular diagnostics.
- Molecular pathology of metabolic diseases.
- Nutrition, signal transduction and gene expression.

PREVIOUS KNOWLEDGE

RELATIONSHIP TO OTHER SUBJECTS OF THE SAME DEGREE

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

OTHER REQUIREMENTS

Before initiating the course in Molecular Pathology it is necessary to have assimilated a series of concepts that the student should have acquired in basic subjects studied during the first year courses as well as in the first semester of the second year. In particular, students must have completed the subjects of Biology, Physiology, Biochemistry I and Biochemistry II.

COMPETENCES / LEARNING OUTCOMES

-

Act with autonomy in learning, making informed decisions in different contexts, issuing judgements based on experimentation and analysis, and transferring knowledge to new situations.

Apply scientific knowledge of physiology, physiopathology, nutrition and food to offer dietary planning and advice to both healthy and sick individuals and communities throughout their life cycle.

Collaborate effectively in work teams, assuming responsibilities and leadership roles and contributing to collective improvement and development.

Contribute to the design, development and implementation of solutions that respond to social demands, taking into account the Sustainable Development Goals as a reference.

Demonstrate critical and self-critical thinking in the field of the degree programme, considering aspects such as professional ethics, moral values and the social implications of the different activities carried out.

Develop the ability for well-founded scientific argumentation, and the habit of using clinical terminology as a natural means of communication with other professionals in the field of health.



Develop the capacity to gather and convey information in English at a level equivalent to the B1 level in the Common European Framework of Reference for Languages.

Develop the necessary skills to communicate and inform the patient and/or user about the content and implications of laboratory reports in appropriate terms.

Identify and know how to apply specific biochemical markers appropriate for assessing health status in the population (individual and collective), and for disease prevention in primary and secondary care.

Interpret, in a global and integrated manner, a patient's overall analytics and understand the appropriate analytical strategy for the differential diagnosis of the various conditions.

Know and understand, within the field of the degree programme, gender inequalities in society; integrate different needs and preferences based on sex and gender into the design of solutions and problem solving.

Know and understand the mechanisms responsible for the genetic variability that characterises humans, and its relationship and implications in human pathology and response to pharmacological therapy.

Know how to communicate effectively, both orally and in writing, adapting to the characteristics of the situation and the audience.

Know the analytical techniques of the clinical biochemistry laboratory, and be able to assimilate and incorporate future innovations both in technical aspects and in relation to the clinical usefulness of each new parameter.

Know the influence of nutrition on pathological states and vice versa.

Know the major diseases at the molecular level.

Know the molecular bases and biochemical mechanisms (genetic, structural and/or functional alterations) of disease, as a rational approach to its diagnosis, treatment and prevention, to the identification of new therapeutic targets, and to the selection a

Know the molecular basis for applying dietary treatments to diseases.

Propose creative and innovative solutions to complex situations or problems within the field of knowledge, to respond to diverse professional and social needs.

Reinforce the acquisition of the general competences of the curriculum.

Saber cómo plantearse problemas y utilizar los métodos adecuados para su resolución, siendo capaz de llevar a cabo un razonamiento crítico.

Ser capaz de llevar a cabo una comunicación oral o escrita.

Ser capaz de trabajar en equipo y de organizar y planificar actividades.

Skills in analysis and synthesis.

Understand the basis and usefulness of the different biochemical markers and functional tests currently applicable to clinical diagnosis, prognosis, disease progression and therapeutic monitoring.



Understand the epigenetic regulation of nutrients.

DESCRIPTION OF CONTENTS

1. Molecular basis of disease.

General concepts. Genes and chromosomes. Flow of genetic information. Mutations. Genes, environment and disease. Categories of genetic diseases.

2. Molecular diagnostic techniques and treatment of genetic diseases

Tools and methods for detecting mutations. Examples of application of diagnostic techniques. Preimplantation diagnosis. Prenatal diagnosis. Postnatal diagnosis. Treatment of clinical phenotype. Treatment directed to the metabolic/protein defect phenotype. Gene therapy. Therapeutic potential of stem cells.

3. Disorders of carbohydrate metabolism.

Metabolism of fructose: essential fructosuria and hereditary fructose intolerance. Metabolism of galactose, galactosemia. Impaired glucose 6-phosphate dehydrogenase. Disorders of glycogen metabolism: glycogen storage disease.

4. Diabetes mellitus.

Definition. Biosynthesis and secretion of insulin. Intracellular effects of insulin. Carbohydrate homeostasis. Classification: diabetes mellitus type I, diabetes mellitus type II, other specific types, gestational diabetes. Diagnosis. Metabolic abnormalities in diabetes. Complications. Control of the diabetic patient

5. Hypoglycemia. Metabolic syndrome

Hypoglycemia. Definition. Regulatory responses. Causes. Classification. Biochemical diagnosis. Definition, epidemiology and biochemical profile. Metabolic syndrome. Diagnostic criteria. Treatment.

6. Metabolism of plasma lipoproteins: dyslipoproteinemias. Hypercholesterolemia and atherosclerosis.

Characteristics of plasma lipoproteins. Lipoprotein metabolism. Alterations in lipoprotein metabolism: dyslipoproteinemias. Diagnosis. Hypercholesterolemia and atherosclerosis. Dyslipoproteinemias



treatment. Dietary factors in the regulation of cholesterol and triglycerides.

7. Disorders of amino acid metabolism

Disorders of amino acid catabolism. Hyperphenylalaninemia and phenylketonuria. Hyperhomocysteinemia and homocystinuria. Alkaptonuria. Disorders of urea cycle. Nutritional treatments

8. Disorders of purine metabolism

Metabolic destinations and overview of the synthesis of nucleotides. Synthesis "de novo" and salvage pathways. Degradation of nucleotides. Disorders of pyrimidine nucleotides metabolism: hereditary orotic aciduria. Disorders of purine nucleotides metabolism: immunodeficiencies. Alterations of the salvage pathway: Lesch-Nyhan síndrome. Hyperuricemia and gout.

9. Disorders of heme metabolism

Biosynthesis of heme. Porphyrrias: congenital erythropoietic porphyria and acute intermittent porphyria. Degradation of heme. Hyperbilirubinemia and jaundice. Jaundice: pre-hepatic (hemolytic) and post-hepatic (obstructive). Biochemical and clinical aspects.

10. Molecular pathology of iron metabolism

Importance of iron in humans. Metabolism and regulation associated genetic pathologies: Hemochromatosis. Nutritional anemias.

11. Molecular pathology membrane transport: Cystic fibrosis

Molecular basis of cystic fibrosis pathology associated model membrane transport. Classification of mutations. Clinical phenotypes. Nutritional interest of phenotypes with pancreatic insufficiency. Diagnosis and treatment.

12. Molecular basis of celiac disease

Definition. Clinical manifestations. Etiopathogenesis. Diagnosis. Treatment.

13. Molecular bases of alcoholic disease.

Ethanol metabolism. Ethanol toxicity. Biochemical markers and associated pathologies.



14. Regulation of gene expression by nutrients

Introduction. Concepts of nutrigenomics, nutrigenetics and nutriepigenetics. Regulation of gene expression by carbohydrates. Regulation of gene expression by lipids. Regulation of gene expression by amino acids and other nitrogen-containing compounds. Regulation of gene expression by vitamins and minerals. Regulation of gene expression by other food components. Nutritional influences on epigenetic patterns, gene expression and phenotypes.

WORKLOAD

PRESENCIAL ACTIVITIES

Activity	Hours
Tutorials	2,00
Theory	38,00
Seminar	2,00
Total hours	42,00

NON PRESENCIAL ACTIVITIES

Activity	Hours
Attendance at other activities	0,00
Individual or group project	12,00
Independent study and work	29,50
Preparation of lessons	26,00
Preparation for assessment activities	0,00
Resolution of case studies	0,00
Total hours	67,50

TEACHING METHODOLOGY

The course is structured as follows:

Lectures: consists of 38 sessions of one hour in which they will be presented with the concepts necessary to enable students to learn each of the concepts. These concepts will be reinforced with other proposed activities, including individual study, the resolution of problems, preparation of seminars by students and tutorials as well as attendance at both.

Specialized group Tutorials: There will be 2 compulsory sessions of one hour, in groups of 16 students, distributed throughout the semester to cover the different thematic blocks of the course. These sessions reinforce the concepts presented in the lectures and encourage the active participation of students. To do this, the teacher will pose questions to be discussed during the session.

Coordinated Seminars: Presentation of the seminar will be compulsory and cover topics proposed by the professor. The presentations will be organized in groups of students. Each of these groups must submit a written report on the proposed topic, including references used for the preparation



thereof. The seminar will be public and may employ any means of presentation that the group members see fit. The presentation will be followed by discussion among the audience, moderated by the professor.

The seminars will be managed by the rules of the degree of Human Nutrition and Dietetics and DG in Pharmacy-Human Nutrition and Dietetics, available in the faculty web.

EVALUATION

The knowledge gained by the student will be assessed by a final evaluation. The final score is 100, broken down into:

- **Evaluation of the theoretical knowledge:** There will be an exam consisting of any combination of multiple-choice and/or short answer questions. The score for this exam is 80.
- **Evaluation of the seminars:** The preparation and presentation of the seminar is mandatory for the student. It will be graded by 10 in the final score. The student's ability to obtain information from bibliographic sources, prepare a team work, present it in public and discuss different aspects of it with the classmates and the teacher will be evaluated.
- **Written assignments:** During the semester, the student will make written assignments on topics related to the content of the course, subjects of current interest in biochemistry, etc., which will contribute 10% to the final score.

It will be necessary to pass each of the sections separately in order to pass the course. It will be necessary to obtain a minimum of 40 in the theoretical exam and 5 in the seminar. If the student does not reach the minimum in the theoretical exam, the final score will not include the score of the seminar and the written assignments.

Attendance at tutorials and seminars is mandatory (except for students who did not pass the course) and, therefore, NOT RECOVERABLE, in accordance with the provisions of Article 6.5 of the Regulation of Evaluation and Qualification of the UV for Bachelor and Master degrees. If it is not possible to attend any of these activities for justified reasons, it must be communicated in advance. In this way, the person in charge of the subject may assign the student a session in another group. Non-attendance could result in a 10% penalty in the final score.

Evidence of copying or plagiarism in any of the assessable tasks will result in failure to pass the subject and in appropriate disciplinary action being taken. Please note that, in accordance with article 13. d) of the Statute of the University Student (RD 1791/2010, of 30 December), it is the duty of students to refrain from using or participating in dishonest means in assessment tests, assignments or university official documents. In the event of fraudulent practices, the "**Action Protocol for fraudulent practices at the University of Valencia**" will be applied (ACGUV 123/2020): <https://www.uv.es/sgeneral/Protocols/C83sp.pdf>

REFERENCES



- Devlin TM. Bioquímica: libro de texto con aplicaciones clínicas. 4ª ed. Ed. Reverté, Barcelona, 2016.
- Gil A. Tratado de Nutrición. Tomos I, II y V. Ed. Panamericana. 3ª ed. 2017.
- González de Buitrago JM, Medina-Jiménez JM. Patología Molecular. McGraw-Hill Interamericana. 2001.
- González Hernández A. Principios de Bioquímica Clínica y Patología Molecular. Ed. Elsevier 3ª ed. 2019.
- Kaplan LA, Pesce AJ Kazmierczak Sc (eds.) Clinical chemistry. Theory analysis, correlation. 5ª ed. Mosby, 2009.
- Jameson JL, Fauci AS, Kasper DL., et al. Harrison: Principios de Medicina Interna. 21ª ed. McGraw-Hill, 2023.
- Lieberman M, Marks AD. Basic medical biochemistry. A clinical approach. 4ª ed. Lippincott Williams & Wilkins, 2012.
- Marshall WJ, Bangert SK. Clinical Chemistry. 9ª ed. Elsevier, 2020.
- McPherson, R.A. and Pincus, M.R. .Henry`s Clinical diagnosis and management by laboratory methods.24th ed. Elsevier, 2022.
- Rifai, N., Horvath, A.R and Witwer, C.T. (eds.) Tietz textbook of clinical chemistry and molecular diagnostics. 6 ed. Elsevier. 20178.
- Rosenthal MD, Glew RH. Medical Biochemistry. Human metabolism in health and disease. John Wiley & Sons, Inc. 2009.
- Smith C., et al. Bioquímica básica de Marks. Un enfoque clínico. 2ª ed. McGraw-Hill Interamericana, 2006.
- Lodish H., et al. Biología celular y molecular. 7ª ed. Editorial Médica Panamericana, 2021.
- Nelson DL, and Cox MM. Lehninger. Principios de Bioquímica. 7ª ed. Ediciones Omega, Barcelona, 2018.
- Voet D, Voet JG, Pratt CW. Fundamentos de bioquímica. La vida a nivel molecular. 4ª ed. Ed. Panamericana, Madrid, 2021.