VNIVERSITAT DE VALÈNCIA

Course Guide
33961 Molecular Pathology

COURSE DATA

Data Subject	
Code	33961
Name	Molecular Pathology
Cycle	Grade
ECTS Credits	4.5
Academic year	2022 - 2023

Study (s)

Degree	Center	Acad. year	Period
1205 - Degree in Human Nutrition and Dietetics	Faculty of Pharmacy and Food Sciences	2	Second term
1211 - Double Degree in Pharmacy and Human Nutrition and Dietetics	Faculty of Pharmacy and Food Sciences	5	Second term

Subject-matter

Degree	Subject-matter	Character
1205 - Degree in Human Nutrition and Dietetics	21 - Molecular pathology and physiopathology	Obligatory
1211 - Double Degree in Pharmacy and Human Nutrition and Dietetics	1 - Asignaturas obligatorias del PDG Farmacia-Nutrición Humana y Dietética	Obligatory

Coordination

Name	Department
GUASCH AGUILAR, ROSA	30 - Biochemistry and Molecular Biology
MIRALLES FERNANDEZ, VICENTE	30 - Biochemistry and Molecular Biology
MURGUI FAUBEL, MARIA AMELIA	30 - Biochemistry and Molecular Biology

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.

- The molecular basis of cancer.

- 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 (RD 1393/2007) // LEARNING OUTCOMES (RD 822/2021)

1205 - Degree in Human Nutrition and Dietetics

- 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.
- Skills in analysis and synthesis.
- Saber cómo plantearse problemas y utilizar los métodos adecuados para su resolución, siendo capaz de llevar a cabo un razonamiento crítico.
- Ser capaz de trabajar en equipo y de organizar y planificar actividades.
- Ser capaz de llevar a cabo una comunicación oral o escrita.
- 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.

- Know the major diseases at the molecular level.
- Know the molecular basis for applying dietary treatments to diseases.
- Understand the epigenetic regulation of nutrients.
- Know the influence of nutrition on pathological states and vice versa.

LEARNING OUTCOMES (RD 1393/2007) // NO CONTENT (RD 822/2021)

Learning outcomes to be achieved with the Molecular Pathology course can be summarized as follows:

- Learn and understand human genetic variability, its biological significance, and its relationship and involvement in human pathology and therapeutic response.

- To understand the molecular basis and the biochemical mechanism (genetic, structural and / or functional alterations) of certain diseases, as a rational approach to diagnosis, treatment and prevention.

- To learn the main techniques used in the diagnosis and treatment of certain diseases.

- Understand how different food components, both nutrients and other non-nutrient chemicals, affecting the health of individuals through the alteration of the structure and expression of genes.

- Understand how the genetic variants of individuals influence the metabolic utilization of nutrients.

- Learn the most appropriate nutritional guidelines for the treatment and prevention of certain diseases.

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 profileMetabolic 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. Porphyrias: 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. Calcium and phosphate metabolism. Molecular basis of genetic and nutritional bone disease.

Calcium and phosphate metabolism. Nutritional interest. Bone remodeling. Molecular pathology of genetic bone diseases (Ostegenesis imperfecta, osteopetrosis) and nutrition (rickets, osteomalacia, osteopenia and osteoporosis)

14. Regulation of gene expression by nutrients

Introduction. Concepts of nutrigenomics, nutrigenetics and nutriepigenética. 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

ACTIVITY	Hours	% To be attended
Theory classes	38,00	100
Tutorials	2,00	100
Seminars	2,00	100
Development of group work	6,00	0
Development of individual work	6,00	0
Study and independent work	29,50	0
Preparing lectures	26,00	0
TOTAL	**109,50**	

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 minimun in the theoretical exam, the final score wil 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). Non-attendance could result in a 10% penalty in the final score.

REFERENCES

Basic

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Additional

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