

**COURSE DATA****Data Subject**

Code	33961
Name	Molecular Pathology
Cycle	Grade
ECTS Credits	4.5
Academic year	2018 - 2019

Study (s)

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

OUTCOMES

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

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 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

Disorders of pyrimidine nucleotides metabolism: hereditary orotic aciduria. Disorders of purine nucleotides metabolism: immunodeficiencies. Alterations of the salvage pathway: Lesch-Nyhan syndrome. Hyperuricemia and gout.

10. Molecular pathology of iron metabolism.

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

11. 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 (Osteogenesis imperfecta, osteopetrosis) and nutrition (rickets, osteomalacia, osteopenia and osteoporosis)

**12. 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.

13. Molecular basis of celiac disease

Definition. Clinical manifestations. Etiopathogenesis. Diagnosis. Treatment.

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.

EVALUATION

The assessment of learning of knowledge and skills gained by the students will be done with a final assessment objective. The maximum rating is 100 points, broken down into:

- **Final exam theoretical lections:** the exam will consist of short questions, developmental and / or multiple response questions. The maximum value of the final exam is 80 points.

Evaluation of the seminars: the preparation and presentation of seminars is compulsory for the student. Be assessed with 10 points in the final grade of the course. Assess the student's ability to extract information from literature sources, prepare a team, expose it publicly and discuss with peers and the teacher different aspects.

- **Evaluation of the tutorials:** It directly assess, assisting them, he proposed tasks and resolving issues about mentoring wonder along the final exam. All this will lead up to 10 points to the final grade for the course.

To pass the course requires passing the theory exam and 40 points to 80 in that review concepts be additional seminars and tutorials. This final note exceed 50 points out of 100 to pass the course.

The note will be retained tutoring during the academic year in which they were made.

REFERENCES

Basic

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- Marshall WJ, Bangert SK Clinical Chemistry 5^a ed. Elsevier Health Science, 2006.
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Additional

- Lodish H., et al. Biología celular y molecular 5^a ed. Editorial Médica Panamericana, 2005.
- Nelson DL, Cox MM. Lehninger. Principios de Bioquímica 5^a ed. Ediciones Omega, Barcelona, 2009.
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