

# COURSE DATA

| Data Subject                |                                   |   |                      |
|-----------------------------|-----------------------------------|---|----------------------|
| Code                        | 34496                             |   |                      |
| Name                        | Molecular principles of pathology |   |                      |
| Cycle                       | Grade                             |   |                      |
| ECTS Credits                | 4.5                               |   |                      |
| Academic year               | 2020 - 2021                       |   |                      |
|                             |                                   |   |                      |
| Study (s)                   |                                   |   |                      |
| Degree                      |                                   | Center                                  | Acad. Period<br>year |
| 1204 - Degree in Medicine   |                                   | Faculty of Medicine and Odon            | tology 4 First term  |
| Subject-matter              |                                   |   |                      |
| Degree                      | 486 584                           | Subject-matter                          | Character            |
| 1204 - Degree in Medicine   |                                   | 18 - Optional subjects                  | Optional             |
| Coordination                |                                   |   |                      |
| Name                        |                                   | Department                              |                      |
| DONATO MARTIN, MARIA TERESA |                                   | 30 - Biochemistry and Molecular Biology |                      |
| SAEZ TORMO, GUILLERMO       |                                   | 30 - Biochemistry and Molecular Biology |                      |

## 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.

## OUTCOMES

#### 1204 - Degree in Medicine

- 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.
- 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.
- Proper organisation and planning of the workload and timing in professional activities.
- Team-working skills and engaging with other people in the same line of work or different.
- Criticism and self-criticism skills.
- Capacity for communicating with professional circles from other domains.
- Acknowledge diversity and multiculturality.
- Consideration of ethics as a fundamental value in the professional practise.
- Working capacity to function in an international context.

# LEARNING OUTCOMES

Students will acquire knowledge on:

- Biochemical and molecular bases of pathologies.
- Disorders of the cellular metabolism and metabolic interactions of tissue.
- The link between clinical manifestations and analytical data.

# **DESCRIPTION OF CONTENTS**



## Vniver§itatöt d'València

## 1. THEORETICAL LESSONS.

## Clinical biochemistry and molecular pathology. The molecular basis of diseases (1 h)

The notion of clinical biochemistry. Its origins and its clinical applications. Molecular pathology as an area of knowledge. Its origin and action lines. The concept of molecular disease. Molecular level in medical conditions. Genetic variability. Types of genetic disorders.

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

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 (2 h)

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 (2 h)

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 (2 h)

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 (2 h)

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 (2 h)

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. Measurement of the Glomerular Filtration Rate. Lightening of plasma creatinine.



## 8. Diagnosis of cardiovascular diseases (2 h)

Biochemistry of the heart muscle and control of the coronary flow. Ethiopathogenia of heart attack. Biochemical markers of interest and their interpretation.

## 9. Tumour markers (2 h)

Cell transformation markers: oncogenes and tumour suppressor genes. Protein tumour markers.

### **10. PRACTICAL LESSONS**

#### CLINICAL PRACTISE.

Practises in laboratories for clinical diagnosis.

Stay in the laboratory of clinical routine and urgent biochemistry, and follow-up of the whole analytical process. 4 hours.

### SEMINARS

1. Omics (Genomics, Transcriptomics, Proteomics, and Metabolomics): basis and their application in clinical diagnosis (2 h).

2. Aminoacidopathies (1 h).

3. Analytical determinations for the measurement of nitrogenous metabolism (nitrogen balance, nutritional status, metabolism of purines, and pyrimidines) (1 h).

4. Biochemical determinations in other biological liquids (urine, faeces, cerebrospinal fluid, etc.) (2 h).

5. Laboratory tests for the study of immunological disorders (hypersensitivities, autoimmunity and immunodeficiencies) (2 h).

6. La toma de muestras y las fases de realización del examen clínico (2 h)

7. La organización del laboratorio analítico en un hospital; control de calidad, errores e interferencias analíticas; seguridad (2 h)

8, 9 and 10. Presentation and discussion on clinical case studies (6 h)

FORMAL TUTORIALS



## Vniver§itatö́dValència

# WORKLOAD

| ACTIVITY                             | Hours  | % To be attended |
|--------------------------------------|--------|------------------|
| Theory classes                       | 19,00  | 100              |
| Seminars                             | 18,00  | 100              |
| Tutorials                            | 4,00   | 100              |
| Clinical practice                    | 4,02   | 100              |
| Development of group work            | 24,00  | 0                |
| Study and independent work           | 31,50  | 0                |
| Readings supplementary material      | 6,00   | 0                |
| Preparation of evaluation activities | 6,00   | 0                |
| TOTAL                                | 112,52 |                  |

# **TEACHING METHODOLOGY**

In theoretical lessons, professors will present the most important concepts and content through master classes in a structured way, in order to help students develop several skills and meet the objectives. Students' participation in the class should be encouraged and they will have access to the didactic material professors may have used through the electronic platform *Aula Virtual*, if they consider it to be appropriate.

Practise in the classroom: seminaries. In small groups, professors will present deeply specialised themes, case studies, management of bibliography, current issues... Team work is encouraged, as well as oral presentation skills, which could be interpreted as 'cooperative learning'.

**Clinical practises**: stays in clinics students have to complete in healthcare services of several university hospitals, primary healthcare centres, mental health centres, and public health areas, as well as learning how to perform and anamnesis and a basic clinical exploration, with a first contact with patients, which will be supervised by a professor.

**Formal tutorials**: they will be done individually or in small groups. Professors will do a follow-up on students' learning process. These will be useful to solve doubts which may arise throughout the different formative activities.

## **EVALUATION**

A written test will be carried out that will aim to assess the acquisition of knowledge of the different contents of course: the theory program, seminars and practices. The written test will consist of short questions and multiple choice questions with 4 response options. In the last ones mentioned, each wrong answer will penalise 25% of a correct answer, and blank answers do not penalise. This **test** will provide **90% of the final score: 50% corresponding to theoretical contents and 40% practical contents** (seminars and practices).



The student enrolled for the first time must attend at least 80% of the practical activities. Attendance of the clinical laboratory practice will be compulsory.

**Evaluation of formal tutorials (10% of the final mark)**: Continuous assessment of participation and work students have done in their groups.

The subject is passed with an overall score of 5. Passing both the theoretical and the practical parts as independent sections is not necessary in order to pass this subject.

In order to access to an advance on the call of this subject, it is a requirement that the student has coursed all his/her practices.

# REFERENCES

## Basic

- González Hernández, A.: Principios de bioquímica clínica y patología molecular. Elsevier España SL 2010.
- Fuentes Arderiu, X. et al.: Bioquímica clínica y patología molecular. Ed. Reverté, 1998.
- Gaw, A.: Bioquímica clínica: texto ilustrado en color. Elsevier, 2001.

## Additional

- González de Buitrago, J.M. et al.: Bioquímica clínica. McGraw-Hill Interamericana, 1998
- González de Buitrago, J.M. et al.: Patología molecular. McGraw-Hill Interamericana, 2001
- Baynes, J.W.: Bioquímica médica. Elsevier 2006
- 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

# ADDENDUM COVID-19

This addendum will only be activated if the health situation requires so and with the prior agreement of the Governing Council

Siguiendo las recomendaciones del Ministerio, la Consellería y el Rectorado de nuestra Universidad, para el período de la "nueva normalidad", la organización de la docencia para el primer cuatrimestre del curso 2020-21, seguirá un modelo híbrido, donde tanto la docencia teórica como práctica se ajustará a los horarios aprobados por la CAT pero siguiendo un modelo de Presencialidad / No presencialidad en la medida en que las circunstancias sanitarias y la normativa lo permitan y teniendo en cuenta el aforo de las aulas y laboratorios docentes. Se procurará la máxima presencialidad posible y la modalidad no presencial se podrá realizar mediante videoconferencia cuando el número de estudiantes supere el coeficiente de ocupación requerido por las medidas sanitarias. De manera rotatoria y equilibrada los estudiantes que no puedan entrar en las aulas por las limitaciones de aforo asistirán a las clases de manera



no presencial mediante la transmisión de las mismas de manera síncrona/asíncrona via "on line".

