

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

Code: 46499
Name: Pharmacogenetics
Cycle: Master's Degree
ECTS Credits: 3
Academic year: 2025-26

STUDY (S)

Degree	Center	Acad. year	Period
2254 - Master's Degree in Molecular Approaches in Health Sciences	Facultat de Medicina i Odontologia	1	Second quarter

SUBJECT-MATTER

Degree	Subject-matter	Character
2254 - Master's Degree in Molecular Approaches in Health Sciences	Biotransformation, metabolism of drugs and xenobiotics	COMPULSORY

COORDINATION

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DONATO MARTIN MARIA TERESA

O'CONNOR BLASCO JOSE ENRIQUE

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

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

OTHER REQUIREMENTS**COMPETENCES / LEARNING OUTCOMES**

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Aprendizaje, manejo y presentación de informes y trabajos en exposición pública de las aplicaciones



biomédicas de los conceptos farmacogenéticos en las distintas terapias actuales, usando como vehículo la lengua inglesa.

Comprender la investigación básica y clínica de la medicina personalizada.

Conocer en profundidad y comprender la organización a nivel molecular de células, sistemas y procesos de relevancia en las Ciencias de la Salud.

Conocer en profundidad y comprender las metodologías de investigación básica aplicables a las Ciencias de la Salud.

Students should apply acquired knowledge to solve problems in unfamiliar contexts within their field of study, including multidisciplinary scenarios.

Students should be able to integrate knowledge and address the complexity of making informed judgments based on incomplete or limited information, including reflections on the social and ethical responsibilities associated with the application of their knowledge and judgments.

Students should communicate conclusions and underlying knowledge clearly and unambiguously to both specialized and non-specialized audiences.

Students should demonstrate self-directed learning skills for continued academic growth.

Students should possess and understand foundational knowledge that enables original thinking and research in the field.

Tener capacidad de analizar y sintetizar un problema.

Tener capacidad de comunicación oral y escrita en una segunda lengua científica.

Tener capacidad de desarrollar un trabajo interdisciplinar.

Tener capacidad de localizar información.

Tener capacidad de trabajar en equipo

DESCRIPTION OF CONTENTS

1. Introduction to Pharmacogenetics and Pharmacogenomics

Pharmacogenetics vs. Pharmacogenomics: historical evolution, field of study and experimental approach. Specific objectives - clinical perspective and pharmacological R&D.

Fundamentals of human genetic variability: SNPs, Indels and CNVs. Functional consequences of genetic polymorphism genotype-phenotype relationships. Most relevant pharmacogenes (PK/PD and ADRs) and



2. The Human Genetic Variability and the Pharmacogenes

databases of interest. The implementation of pharmacogenetics in clinical practice.

3. Molecular Methods in Pharmacogenetics

Molecular technologies to determine the genotype, applicable to the detection of polymorphisms. DNA sequencing: Capillary and Massive (NGS). Amplicons for detection of SNPs. multiplexed PCRs. Taqman trials. Biomarkers

4. Pharmacogenetics of Drug Metabolism (I): Phase I Drug-metabolizing enzymes

Interindividual variability: causes. Molecular basis of the CYP2D6, CYP2C9, CYP1A1 and CYP2E1 polymorphism. Other CYPs. Other Phase 1 enzymes

5. Pharmacogenetics of Drug Metabolism (II): Phase II Drug-metabolizing enzymes

Glutathione-S-transferases. N-acetyltransferases. Methylation enzymes, conjugation with glucuronic acid, flavinmonooxygenases, and others.

6. Pharmacogenetics of Drug Transporters

Drug transporters and xenobiotics. Classification and functions. Drug resistance mechanisms. Clinical consequences of polymorphisms in drug transporters. Techniques for the study of drug transporters.

7. Pharmacogenetics of Drug Adverse Reactions

Molecular basis of the different types of ADRs. Genetic predisposition. Strategies for the design of safe drugs.

8. Clinical Implications of Pharmacogenetics (I): Gene Polymorphisms in Hemostasis

Impact of gene polymorphisms in platelet function and antiaggregant therapy



9. Clinical Implications of Pharmacogenetics (II): From Sequence to Precision Medicine

Future clinical applications: Allelic imputation, Phenotypic imputation, Validation of functional evidence, Validation of associations with adverse drug reactions for clinical practice guidelines. Anticoagulants. Cancer. Antidepressants.

10. Clinical Implications of Pharmacogenetics (III): Pharmacogenetics in Translational Research and Clinical Practice

WORKLOAD

PRESENCIAL ACTIVITIES

Activity	Hours
Theory	20,00
Group work	10,00
Total hours	30,00

NON PRESENCIAL ACTIVITIES

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

TEACHING METHODOLOGY

EVALUATION

The evaluation of student learning will be carried out by evaluating the following sections:

1. Fulfillment of a series of tasks proposed by the teachers of the subject and that will be available in the Virtual Classroom. The tasks will imply the development of questions of a practical nature or clinical relevance. Students will receive the necessary instructions and bibliography to solve the tasks, which will be graded at the end of the semester. From this section you can get up to 90% in the final grade of the subject.

2. Student interest in the subject, expressed as her participation in organized discussions, attendance at personal tutorials and/or any other type of activity carried out by the student in relation to the subject. Up to



10% of these concepts can be obtained in the final grade of the subject.

REFERENCES

- Pharmacogenomics. Eds.: W. Karlow, U.A. Meyer & R.F. Tyndale, Taylor & Francis, New York, 2005
Rapid Review Pharmacology 3^a ed. Eds.: T.L. Pazdernik & L. Kerecsen, Mosby 2010
Handbook of drug metabolism. 2^a ed. P.G. Pearson & L.C. Wienkers, Informa Healthcare USA, 2008
Cytochrome P450. Structure, mechanism and biochemistry. 3^a ed. Ed.: P.R. Ortiz de Montellano, Kluwer Academic/Plenum Press, New York, 2005
Complementarias