SUBJECT TEACHING GUIDE

G1944 - Applied Genetics and Personalized Medicine

Degree in Biomedical Sciences

Academic year 2023-2024

1. IDENTIFYING DATA					
Degree	Degree in Biomedical Sciences			Type and Year	Optional. Year 4
Faculty	Faculty of Medicine				
Discipline					
Course unit title and code	G1944 - Applied Genetics and Personalized Medicine				
Number of ECTS credits allocated	6	Term		Semester based (1)	
Web					
Language of instruction	Spanish	English Friendly	No	Mode of delivery	Face-to-face

Department	DPTO. MEDICINA Y PSIQUIATRIA
Name of lecturer	JOSE ANTONIO RIANCHO MORAL
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Other lecturers	ANA MARIA FONTALBA ROMERO DOMINGO GONZALEZ-LAMUÑO LEGUINA MARIA TERESA MARTINEZ MERINO ANA SANTURTUN ZARRABEITIA ANDREA SARIEGO JAMARDO ÁLVARO DEL REAL BOLT MARÍA ESTHER ONECHA DE LA FUENTE MONICA GARCIA CASTRO NURIA PUENTE RUIZ NEREA PAZ GANDIAGA

3.1 LEARNING OUTCOMES

- 1. List the genomic alterations involved in common genetically based disorders
2. Identify the diagnostic methodologies available to evaluate genetically based disorders at the pre- and postnatal level.
3. Select the most effective diagnostic procedures for the main syndromes of genetic origin
4. Explore the databases and extract useful information for the interpretation of the anomalies detected in the genetic tests
5. Identify genomic and other biomarkers that help individualize and personalize treatments for non-tumor disorders.
6. Solve problems of human identification and family relationships through the use of genetic tests
7. Identify legal and bioethical problems associated with genetic tests applied in clinical and research and resolve them appropriately.
8. Apply the results of genetic tests for genetic counseling
9. Design genetically based research studies and critically analyze the literature
10. Assess family history and document it in a standardized manner

4. OBJECTIVES

To provide students with knowledge and skills so that they know the bases of
1. Genome and epigenome as causes of disease and modifiers of the response to external agents
2. Genomic and non-genomic markers in Personalized Medicine
3. Analysis techniques in pre and postnatal Clinical Genetics
4. Research methodology and critical analysis of genetic studies
5. Approach to the main genetic syndromes
6. Genetic procedures in human identification
7. Ethical and legal aspects of genetic studies
8. Genetic counseling
9. Application of personalized Medicine to non-tumor processes
10. Gene therapy

And that they develop the necessary skills to

1. Select the analysis techniques in pre and postnatal Clinical Genetics
2. Critically analyze genetic studies and design new studies
3. Interpret DNA studies in identification and parentage
4. Start genetic counseling
5. Develop and interpret family pedigrees
6. Interpret studies of chromosomal alterations
7. Explore bioinformatics bases
8. Elucidate the functional impact of genetic variants
9. Prioritize genetic variants

6. COURSE ORGANIZATION	
	CONTENTS
1	LECTURES 1 Genome and epigenome as causes of disease. 2 Mutation analysis techniques 3 Chromosomal analysis techniques 4 Epigenome and disease. Analysis Techniques 5 Biochemical and Imaging Biomarkers 6 Preimplantation and prenatal diagnosis 7 Hereditary disorders of metabolism. Newborn screening 8 Useful Databases on Genetics and PM 9 Growth disturbances, localized and generalized 10 Neurodevelopmental disorders. Other neurological diseases 11 Endocrine-metabolic diseases 12 Skeletal and connective tissue disease 13 Disorders of the Urinary and digestive systems 14 Heart and Lung Diseases 15 Summary: Approach to the study of the main genetically-based syndromes and diseases 16 Critical Analysis of Studies Using Genetic/Epigenetic Information 17 Ethical and legal aspects of genetic tests and studies. Biobanks 18 Genetic counseling 19 Complex hereditary diseases. polygenic risk 20 Forensic genetics. Identification and parentage 21 Gene therapy 22 Somatic mutations in non-tumor pathology
2	SEMINARS AND PRACTICE EXERCISES 1 Family trees: elaboration and interpretation 2.3 Forensic Genetics: identification, parentage, mixtures and chimerism 4 Cases of genetic counseling 5 Bioinformatic databases 6.7 Analysis of individual variants 8.9 Analysis of Variant Listings 10.11 Analysis of arrays 12 Critical Analysis of Studies Using Genetic/Epigenetic Information 13 Preparation of a research project
3	Visits to clinical labs
4	Computer practice
5	Tutorship
6	Evaluation
7	Personal work
8	Group work

7. ASSESSMENT METHODS AND CRITERIA

Description	Type	Final Eval.	Reassessn	%
Continuing practical activities	Work	No	No	40,00
Written multiple-choice questions	Written exam	Yes	Yes	60,00
TOTAL				100,00

Observations

A score of 50% at the paper exam is needed to pass.
Students who pass that score will get a final score by adding the paper exam and practical activities weighted scores,

Observations for part-time students

General criteria adapted to part-time schedules

8. BIBLIOGRAPHY AND TEACHING MATERIALS

BASIC

-Dhar SU et al. Handbook of clinical adult genetics and genomics. A parctice-based approach. Academic Press. 2020.

- Firth HV y Hurst JA. Clinical genetics and genomics. Oxford University Press. 2017