Universidad de Navarra

Human Molecular Genetics (F.Ciencias)

Teaching guide 2023-24

1. INTRODUCTION

Human Molecular Genetics deals with the structure and function of the human genome, and the many ways in which genetic and genomic alterations lead to human disease. It is a core course of the Degree in Biochemistry and an optional course of the Degree in Biology.

This course carries 6 ECTS (150 hours of personal work by the student). It is taught during the **first semester** by Prof. Francisco Javier Novo Villaverde

Lecture times 2023-2024: Tuesday 11:00-12:00 - Wednesday 10.00-11.00 - Thursday 09:00-10:00.

ROOM 12 in Biblioteca de Ciencias.

This course follows a **Team-Based Learning** format. Students are randomly assigned to a team (made up of four members), team members should sit together in the room and answer the questions and exercises during the class, some of them using WOOCLAP. Team performance will be evaluated (see "Evaluation").

To check the **composition of teams and their location in Room 12**, see "Teams" and also the tab "Anuncios".

A **case-competition** will be held in **Lab 0310** (basement of the Biblioteca de Ciencias) from **16.00 to 19.00** according to the following calendar:

- **Group 1:** 23, 24 and 25 October
- **Group 2:** 30 and 31 October, and 2 November
- **Group 3:** 7, 8 and 9 November

Compositions of groups for the Case Competition will be announced in due course.

2. COMPETENCES

Competences.png

3. PROGRAM

Unit 1. The landscape of the human genome

- 1.1 Deciphering the sequence of the human genome.
- 1.2 What do we find buried in the sequence of the human genome?
- 1.3 Repetitive and "junk" DNA. Genomic disorders.
- 1.4 Forensic applications of genomic variation.

Unit 2. The human genome in action

- 2.1 The functional state of chromatin correlates with epigenetic modifications: methylation of DNA; epigenetic modifications of histones.
- 2.2 Beyond the HGP: the ENCODE project; the Roadmap Epigenomics Project.
- 2.3 Chromatin 3D conformation: loops and TADs.
- 2.4 Inactivation of the X chromosome.
- 2.5 Genomic Imprinting.

Unit 3. Towards Genomic Medicine

- 3.1 Finding genetic factors responsible for complex diseases: heritability.
- 3.2 Genome-Wide Association Studies. Missing heritability (high-risk rare variants?).
- 3.3 Towards individual genomes and “Precision Medicine”.
- 3.4 The future of genome editing.

Unit 4. Causes of genetic variation in humans

- 4.1 Variation in DNA: mutation and recombination.
- 4.2 DNA repair mechanisms in humans (and what happens when they don’t work):
- 4.2.1 MMR (STR expansions)
- 4.2.2 BER
- 4.2.3 NER (XP)
- 4.2.4 DSB repair (IG rearrangements)

Unit 5. Mutation as the cause of disease

- 5.1 Pathogenic potential of mutations in coding DNA
- 5.2 Splicing and its regulation
- 5.3 Pathogenic potential of mutations that change splicing patterns
- 5.4 Phenotypic effects of mutations
- 5.4.1 Loss-of-function and gain-of-function mutations
- 5.4.2 Are all pathogenic variants really pathogenic?
- 5.5 Mitochondrial mutations

4. EDUCATIONAL ACTIVITIES

The student will achieve the proposed goals by:

- study of materials provided for each lecture
- case competition
- the preparation of evaluation tests

Distribution of time:

	ECTS	hours

In-class		
Lectures	1.4	36
Case competition	0.4	9
Exams	0.2	5
Subtotal	*2.00*	*50*
Non-presential		
Personal or group study	4.00	100
Subtotal	*4.00*	*100*
TOTAL	**6**	**150**

Case competition

Each team will have to perform molecular diagnosis and genetic counselling in a family with a genetic disease. Over the course of three days (three hours per day), each group will receive some starting information and they will have to process it step-by step, requesting whatever information they need, until completion of the diagnosis and counselling. The last day, each group will subit a report.

5. ASSESSMENT

The **final mark** of the course will be the result of:

- The quizzes and exercises performed by teams during the course, **up to 2 points**.
  - The **case competition** will be evaluated according to individual performance and the final group report, **up to 1 point**.
- The **final test** at the end of the course (a combination of MCQ and short questions) will give **up to 7 points**.

Students with special educational needs: Exceptions will be allowed in Methodology and Evaluation in order to facilitate the achievement of the skills and the goals of this Course.

Students who fail the course will have to take the exam in June; in this case 100% of the mark will be the result of the exam (MCQ test plus short questions).

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6. BIBLIOGRAPHY AND RESOURCES

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HERE you can download the **Lecture Notes** for this course, prepared by Prof. Novo:

Apuntes HMG 2023.pdf

Further reading:

- T. Strachan, A.P. Read: Human Molecular Genetics, 5th edition. Garland Science, 2019. Find it in the library
- Francisco J. Novo: Genética Humana: conceptos, mecanismos y aplicaciones de la Genética en el campo de la Biomedicina. Ed. Pearson 2007 (out of print). Find it in the Library

You can find many **videos** relevant to this course (many of them in Spanish) in my YouTube channel.

It will also be useful to look up the **Glosario of genetic terms** (in Spanish) prepared by Prof. Novo.

7. OFFICE HOURS

Please make an appointment with Dr. Novo by email: fnovo@unav.es

Department of Biochemistry and Genetics

Edificio de Investigación, 3rd Floor (room 3341)