Universidad de Navarra

Human Molecular Genetics (F.Ciencias)

Teaching guide 2025-26

1. COURSE DESCRIPTION

MODULE V: PHYSIOLOGICAL INTEGRATION AND BIOMEDICAL APPLICATIONS

MATTER 5.2 Biomedical Applications

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:

- 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.

To check the **composition of teams**, see "Team composition".

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:** 4-6 November
- **Group 2:** 11-13 November
- **Group 3:** 18-20 November

Compositions of groups for the Case Competition will be announced in due course. Over the course of three days (three hours per day), each group will receive some starting information about a case of a family with a genetic disease, and they will have to process it step-by-step (requesting whatever information they need, analyzing the results from the molecular genetics lab, etc.) until completion of the diagnosis and genetic counseling. On the last day, each group will submit a report.

BIBLIOGRAPHY AND RESOURCES

IMPORTANT NOTICE:

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

Apuntes HMG

Further reading:

- T. Strachan, A.P. Read: **Human Molecular Genetics**, 5th edition. Garland Science, 2020. Find it in the Library
- Tom Strachan and Anneke Lucassen: **Genetics and genomics in medicine.** CRC Press/Taylor & Francis Group, 2023. 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

2. LEARNING OUTCOMES

BQ-C5 Comprender las similitudes y diferencias entre los organismos vivos. Conocer la estructura y función de la célula procariota y eucariota, así como la estructura, variación, función y transmisión del material hereditario

BQ-C6 Conocer la estructura y función de los tejidos, órganos y sistemas animales y humanos. Comprender la regulación e integración de las diversas funciones del organismo para su aplicación en Biomedicina

BQ-C7 Conocer las alteraciones moleculares de los procesos biológicos en situaciones patológicas y los fundamentos de los tratamientos farmacológicos.

BQ-C8 Conocer las bases y las aplicaciones de la tecnología del DNA recombinante, así como las diferentes metodologías instrumentales cuantitativas utilizadas en Bioquímica y Biología Molecular.

BQ-C10 Conocer los principales temas de debate y los retos futuros de la investigación biomédica, su dimensión social y económica, así como sus aplicaciones prácticas.

BQ-H3 Adquirir destreza en la interpretación de las alteraciones moleculares causantes de patología humana y de los resultados de análisis clínicos en sus diferentes modalidades.

BQ-H5 Trabajar en equipo con una adecuada distribución de funciones, sabiendo seleccionar y elegir la metodología de trabajo. Trabajar con sentido de responsabilidad, utilizando los recursos e instalaciones de manera sobria y cuidadosa.

BQ-S4 Pensar de forma integrada y abordar los problemas desde diferentes perspectivas, con creatividad, razonamiento crítico y actitud de servicio. Plantear soluciones positivas, prácticas y factibles desde la perspectiva del conocimiento científico.

3. PROGRAM

Universidad de Navarra

Unit 1. The landscape of the human genome

- Organization of the human genome
- Variable G+C composition in the nuclear genome
- Heterochromatin DNA and Transposon Repeats
- DNA Forensics: Identifying Individuals and Relationships
- The human genome contains many genes of different classes
- The scale of human genetic variation

Unit 2. The human genome in action

- Chromatin accessibility and conformation
- Histones and other DNA-binding proteins
- Regulation by DNA methylation
- X-inactivation, imprinting, and epigenetic memory

Unit 3. Towards Genomic Medicine: identifying susceptibility factors for complex disease

- Investigation of complex disease: epidemiological approaches
- Investigation of complex disease using association
- The limitations of genome-wide association studies
- Towards genomic medicine

Unit 4. The origin of human genetic variation

- Origins of DNA sequence variation
- Endogenous and exogenous sources can cause damage to DNA by altering its chemical structure
- Base mismatch repair (MMR)
- Unrepaired expansions of trinucleotide repeats result in neurological disease
- Base excision repair
- Nucleotide excision repair
- Repair of DNA lesions that affect both DNA strands
- Extraordinary genetic variation in the adaptive immune system

Unit 5. Mutation as the cause of disease

- Molecular pathology
- Loss of function mutations arise in many ways
- Loss of function mutations usually give rise to recessive phenotypes, with some exceptions
- Gain of function mutations are the cause of many genetic diseases with dominant inheritance
- Molecular pathology of mitochondrial disorders

4. EDUCATIONAL ACTIVITIES

PRESENTIAL ACTIVITIES	Hours
Lectures	34

Case Study Competition	9
Evaluation	3
NON-PRESENTIAL ACTIVITIES	
Personal study	65
TOTAL	111

5. EVALUATION

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

- The **case competition** will be evaluated according to individual performance and the final group report, **up to 2 points**.
- The **final exam** at the end of the course will consist of the resolution of a practical exercise, with a combination of multiple-choice (1 point) and short questions (7 points). It will give **up to 8 points**.
- The members of the team that performs best during the course will be rewarded accordingly.

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

STUDENTS WITH SPECIAL NEEDS: Students with special educational needs must contact the Academic Coordination Office of the Faculty of Sciences in advance to obtain the corresponding authorization for any necessary accommodations (for example, additional time for examinations). This authorization must be submitted by the student to the instructor. It is recommended that this process be completed at the beginning of the semester.

NOTICE: Please be reminded that any attempt at **fraud, cheating, plagiarism**, or other forms of academic misconduct constitutes a serious offense, as stipulated in Title IV, "Regulations on Academic Discipline of Students," within the System of Norms on Coexistence at the University of Navarra.

6. EVALUATION OF LEARNING OUTCOMES

	FINAL EXAM	CASE COMPETITION	IN-CLASS ASSESSMENT

BQ-C5	X		
BQ-C6	X		
BQ-C7	X	X	
BQ-C8	X		
BQ-C10	X		X
BQ-H3		X	
BQ-H5		X	
BQ-S4		X	X

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)