Universidad de Navarra

***Genomics and Proteomics (Ing.Gr.20)***
*Guía docente 2026-27*

# PRESENTACIÓN

**Breve descripción**:

This course provides a comprehensive overview of the field of genomics, genetics and proteomics, focusing on the fundamentals of human genetics and genomics, inheritance patterns, functional genomics, technologies utilized in genomics research, genome-wide association studies (GWAS) and expression quantitative trait loci (eQTL) analysis, ethical considerations, legal aspects, and social implications of applied genomics. The course also explores the relationship between genomics and the patient, and dwelves into practical applications of genomics in various fields.

**Titulación (Módulo/Materia)**:

- Ingeniería Biomédica (Bioingeniería/Técnicas high-throghput )

**Detalles**:

- **ECTS**: 4 ECTS
- **Curso, semestre**: 3.º curso, 2.º semestre
- **Carácter**: Obligatorio
- **Idioma**: English

**Profesores de la asignatura**:

- Otaegui Bichot, David/ Profesor Invitado
- Rubio Díaz-Cordovés, Ángel / Profesor Catedrático

# RESULTADOS DE APRENDIZAJE (Competencias)

### INGENIERÍA EN INGENIERÍA BIOMÉDICA

CE8 - Tener una visión conjunta de las funciones celulares y la interacción entre los diferentes “agentes” (proteínas, DNA, RNA) que existen dentro de la célula, así como los algoritmos para buscar estas relaciones.

CE19 - Conocimiento de los algoritmos utilizados en microarrays de expresión génica (tanto estándar como de exones) y en arrays de SNPs.

# PROGRAMA

**1. Fundamentals of Human Genetics and Genomics**

- Importance of the topic
- Central dogma
- Genome structure (chromosomes, genes, cytogenetics -nucleosomes, DNA methylation...)
- Transcription, Translation, Replication
- Alternative splicing.
- Epigenetics
- Gene structure
- Variants:

- Germinal vs Somatic
- Small ones: mutations, polymorphisms, repeats, SNPs
- Chromosomic: amplification, deletion, translocation, inversion, LOH.
- Nomenclature
- Effect of different variants

**2. Inheritance**

- Meiosis
- Linkage desquilibrium
- Population genetics
- Mendelian inheritance
  - Dominant vs recesive
  - Autosomic vs sexual chromosome and mitochondria associated
- Non-Mendelian inheritance.

**3. Functional Genomics**

- RNA expression
- Regulation of gene expression
- Gene Ontology
- Genomic Databases
- In vitro testing
  - Cell cultures
  - CRISPR-Cas9, siRNA
- Clinical Report

**4. Technologies**

- standard PCR and qPCR
- FISH
- Microarrays
- Sequencing (NGS y capilar)
- DNAseq
    - WGS
    - WXS
  - Targetted
  - RNAseq
  - Other applications (ATACseq, bifsulfite sequencing, Chip-seq)
  - Single cell sequencing and spatial transcriptomics.

**5. GWAS and eQTL**

- Concept
- Inference of SNPs (Distance, centimorgans)
- Statistics related to the field (logistic regression, size effect, Manhattan plots, etc.)
- eQTL concept

**6. Ethical, Legal and Social Issues in Applied Genomics. Genomics and the Patient**

**7. Applications**

## ACTIVIDADES FORMATIVAS

Throughout the course, students will engage in lectures, discussions, case studies, and practical exercises to gain a solid foundation in the principles and applications of genomics and genetics. By the end of the course, students will be equipped with the knowledge and skills necessary to understand, critically evaluate, and contribute to the rapidly evolving field of genomics and genetics.

We will have two 1h20m theoretical classes each week. At the beginning of most classes, there will be a short quiz on the contents of the previous topics.

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# EVALUACIÓN

**CONVOCATORIA ORDINARIA**

Grading will be done by weighting the result of the final exam, the assignment and the quizzes:

- The final exam (50%). If the mark in the final exam is below 4.0, the result of the exam will be the final mark. The exam is composed of two parts. It is compulsory to have at least 3.5 in each of the parts.
- The assignment (20%)
- Theoretical and hands-on quizzes (30%) (the worst quiz will not be taken into account).

Assignment:

- It is compulsory
- It will be done in groups of three or four people. One of them will be randomly selected to do the presentation and another one will be selected to answer the questions of the lecturers. All the members of the group will have the same score.

In the final exam and quizzes, no external resources will be allowed.

If for any serious and justified reason, a student cannot attend to one of the quizzes, the result of the quiz will not be included in the final grade. The weights of the rest of the quizzes will be minorated accordingly.

**CONVOCATORIA EXTRAORDINARIA**

- There is no option for assignment. The grade will be 100% the final exam.

# HORARIOS DE ATENCIÓN

Horario de tutoria: Se concretará con el delegado a principio de curso.

**Dr Angel Rubio (arubio@unav.es)**

- Despacho M018. Edificio Miramón. Planta 0.

**Dr. David Otaegui (**dotaegui@gmail.com**)**

# BIBLIOGRAFÍA

This is an evolving topic and most bibliography sources become obsolete in few years.

We will use papers and Internet resources instead.