

**COURSE DATA****Data Subject**

|                      |                             |
|----------------------|-----------------------------|
| <b>Code</b>          | 33140                       |
| <b>Name</b>          | Genetic analysis techniques |
| <b>Cycle</b>         | Grade                       |
| <b>ECTS Credits</b>  | 4.5                         |
| <b>Academic year</b> | 2020 - 2021                 |

**Study (s)**

| <b>Degree</b>                                         | <b>Center</b>                  | <b>Acad. year</b> | <b>Period</b> |
|-------------------------------------------------------|--------------------------------|-------------------|---------------|
| 1109 - Degree in Biochemistry and Biomedical Sciences | Faculty of Biological Sciences | 3                 | Second term   |

**Subject-matter**

| <b>Degree</b>                                         | <b>Subject-matter</b>       | <b>Character</b> |
|-------------------------------------------------------|-----------------------------|------------------|
| 1109 - Degree in Biochemistry and Biomedical Sciences | 10 - Métodos instrumentales | Obligatory       |

**Coordination**

| <b>Name</b>           | <b>Department</b> |
|-----------------------|-------------------|
| FERRE MANZANERO, JUAN | 194 - Genetics    |
| PARICIO ORTIZ, NURIA  | 194 - Genetics    |

**SUMMARY**

The subject of Genetic Analysis Techniques is taught in the third year of the Degree in Biochemistry and Biomedical Sciences (Plan 2009), in the second quarter of the year. This is a compulsory subject. Together with the Genetics-Cytogenetics, Genomics-Genetic Engineering (all mandatory subjects), Genetic Analysis Techniques aims to provide to students the basic knowledge about biological inheritance, as well as the conceptual and methodological tools that enable students to carry out, in a professional work, genetic analysis and genetic modification of organisms.

Students take this course after attending the subjects mentioned above; consequently they already have basic understanding of the biological inheritance and the genome structure and knowledge of molecular tools for genetic analysis.



## PREVIOUS KNOWLEDGE

### Relationship to other subjects of the same degree

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

### Other requirements

## OUTCOMES

### 1101 - Degree in Biochemistry and Biomedical Sciences

- Capacidad para trabajar correctamente en los laboratorios de bioquímica, genética, biología molecular y celular incluyendo seguridad, manipulación, eliminación de residuos y registro anotado de actividades.
- Capacidad para utilizar la instrumentación básica en experimentación molecular y celular.
- Tener una visión integrada de las técnicas y métodos utilizados en biociencias moleculares y biomedicina.
- Capacidad para diseñar experimentos y aproximaciones multidisciplinares para la resolución de problemas concretos.
- Capacidad para presentar, discutir y extraer conclusiones de los resultados de los experimentos científicos.

## LEARNING OUTCOMES

- Understanding and correctly applying the basic concepts of genetic analysis. Thus, the student will be able to:
  1. Know the importance (and value) of the mutants and neutral molecular variability in genetic analysis.
  2. Know the different types of molecular markers used in genetic analysis.
  3. Assign loci to specific chromosomes.
  4. Correlate genetic and cytogenetic maps using chromosomal deletions.
  5. Learn the techniques used in gene mapping in humans and in other organisms.
  6. Perform genetic maps in haploid eukaryotes.
  7. Perform physical maps using restriction enzymes.
  8. Know DNA profiling analysis for identifying individuals and determining relationships
- To encourage student planning effort and independent learning in a continuous form.
- To foster critical view of different aspects of genetic analysis discussed throughout the course, emphasizing the capacity for synthesis and association between different concepts learned.
- Acquisition of ability and skills to design, plan and carry out laboratory experiments.
- Acquisition of ability to extend knowledge through the use of bibliographic sources, both from books, scientific journals and web pages.
- Acquisition of capability to discuss from rational criteria, clearly differentiating what is arguable to what they are made or accepted with scientific evidence.
- Ability to interact with both the teacher and colleagues.



- Ability to build a comprehensive written text
- Acquisition of social and professional awareness about the ethical problems of genetic analysis.
- Professional training: ability and skills to deal with and solve analytic-genetic questions, especially related to gene mapping, molecular markers based linkage and determination of individual identity related to forensic genetics and paternity testing.

## DESCRIPTION OF CONTENTS

### 1. LESSON 1. SEGREGATION AND RECOMBINATION MAPPING IN HAPLOIDS.

Segregation in haploids. Calculating gene-to-centromere distances. Calculating the distance between two loci. Three-point mapping in haploids. Problem solving.

### 2. LESSON 2. COMPLEMENTATION AND DELETION MAPPING.

Complementation test Mapping using deletions in species with giant chromosomes. Combined use of deletions and in situ hybridization for mapping human genes. Problem solving.

### 3. LESSON 3. TYPES OF GENETIC MARKERS.

Importance of biological variability in the genetic analysis. Morphological, biochemical and molecular markers. Identification of a molecular marker linked to a mutant phenotype. Problem solving.

### 4. LESSON 4. GENETIC IDENTITY.

Use of molecular markers to obtain genetic fingerprints. Probability assignment in forensic genetics. Probability assignment in paternity testing. Problem solving.

### 5. LESSON 5. LINEAGE MARKERS.

What are lineage markers. The use of Y-chromosome markers. The use of mitochondrial markers. Problem solving.

### 6. LESSON 6. ASSIGNING LOCI TO SPECIFIC CHROMOSOMES.

Use of linkage group markers in model species. Procedure in species with crossing-over limited to one of the two sexes. Procedure in species with crossing-over in both sexes. Problem solving.

**7. LESSON 7. LINKAGE DETECTION IN HUMANS.**

Linkage analysis in pedigrees. LOD score. Human-mouse somatic cell hybrids. Problem solving.

**8. LESSON 8. OBTAINING RESTRICTION MAPS.**

Restriction maps. The use of probes. Circular maps. Linear maps. Problem solving.

**9. LABORATORY PRACTICALS**

LAB PRACTICAL 1. USE OF MOLECULAR MARKERS. Linkage of a mutant phenotype to a molecular marker using the RAPDs technique.

LAB PRACTICAL 2. SEGREGATION IN HAPLOIDS. Ordered tetrad analysis (estimate the gene-centromere distance in *Sordaria fimicola*).

LAB PRACTICAL 3. GENE LOCALIZATION BY DELETION MAPPING. Precise gene localization using chromosome deletions in *Drosophila melanogaster*.

LAB PRACTICAL 4. GENETIC IDENTITY. Obtaining a genetic fingerprint from saliva and blood DNA, by means of genetic and biochemical markers.

**WORKLOAD**

| ACTIVITY                                     | Hours         | % To be attended |
|----------------------------------------------|---------------|------------------|
| Classroom practices                          | 21,00         | 100              |
| Laboratory practices                         | 18,00         | 100              |
| Tutorials                                    | 3,00          | 100              |
| Theory classes                               | 3,00          | 100              |
| Development of individual work               | 5,00          | 0                |
| Study and independent work                   | 22,00         | 0                |
| Readings supplementary material              | 5,00          | 0                |
| Preparation of evaluation activities         | 16,00         | 0                |
| Preparation of practical classes and problem | 10,00         | 0                |
| Resolution of case studies                   | 9,50          | 0                |
| <b>TOTAL</b>                                 | <b>112,50</b> |                  |

**TEACHING METHODOLOGY**

The following teaching methods will be used to develop this subject:



**1. Theory and problem classes.** A total of 16 sessions of one-hour and a half will be needed to cover this teaching activity in groups of 40 students. The professor will show the most relevant concepts needed to solve the different questions related to each topic. After that several practical problems will be solved.

**2. Laboratory classes.** Five sessions of 3 hours long will be held in groups of 16 students. Attendance is compulsory for laboratory classes.

**3. Group mentoring.** Two sessions of one hour and a half are organized in groups of 16 students. Its aim is to review and discuss the concepts taught during the course. In addition, the final half hour of each session will be devoted to the completion of a written test to evaluate the knowledge acquired by the students. It is intended that these tutorials will stimulate the sustained study of the subject.

**4. Personalized tutoring.** Students will be encouraged to use this resource for advice and discussion with the teacher any topic about the program, the course, or the degree studies.

## EVALUATION

**Application of the acquired concepts to the problem solving:** The evaluation of the concepts worked in the theoretical-practical sessions of problems will be made through a final written exam, which will consist of the presentation of problems and / or questions on any of the aspects dealt with in the classes of problems and tutorials, and two written tests during the group tutorials (about the two parts of the subject). The value of the final test will be 60% of the total, and the value of the two partial tests will be 10%. Both marks will be averaged, weighted (6: 1), only if the mark of the tutorials is higher than that of the final exam (ie, only if it benefits the student).

**Laboratory:** Evaluation of the learning in the laboratory, taking into account the attendance, as well as the presentation of a questionnaire on the results of the lab exercises and the analysis of the same. The value of the laboratory mark will be 30% of the total. The attendance to the laboratory classes is obligatory and essential to pass the subject.

**Portfolio of the student:** In addition, the student can get up to 1.3 extra points in the final score of the problems part with his "portfolio". This consists of having obtained a score of 8.5 or higher in the set of two partial tests in the group tutorials. The application of the portfolio will consist of multiplying by 1.1 the score obtained in the weighted average of the problems section.

Summary of the evaluation system:

### Other considerations:

The final grade results from the sum of the grades achieved in the different sections. To pass the course is necessary to obtain an overall rating equal to or higher than 5 points out of 10, taking into account that the score of theoretical and practical knowledge (upon addition of the "portfolio") and laboratory skills are either, independently, 4,5 or more out of 10.

Students who have not passed the subject in the first call, their marks of the theoretical and practical knowledge will be saved for the second call when they are equal to or higher than 5 points out of 10. However the student can waive these marks performing the corresponding section in the examination of the second call. The score obtained from the laboratory skills is saved for the second call if necessary and for the next year, as well as for subsequent years.





Students who do not assist to the final exam will have NOT EVALUATED in the records.

To apply for an advance of the examination of this course the student must have completed (in any of the previous years) the laboratory classes of this subject.

## REFERENCES

### Basic

- Pierce, B.A. (2009). Genética, un enfoque conceptual. 3ª edición. Ed. Médica Panamericana. ISBN: 978-84-9835-216-0.
- Griffiths, A.J.F., Wessler, S.R., Lewontin, R.C. y Carroll, S.B. (2013). Genética, 9a edición. McGraw-Hill-Interamericana. ISBN: 978-84-481-9090-3
- Klug, W., Cummings, M.R., Spencer C. A. y Palladino, M.A. (2013). Conceptos de Genética. Prentice Hall. (Traducción de la 10ª ed.). ISBN: 978-84-1555-249-9
- Pascual, L. y Moltó, M.D. (1999) Però, què és això de la Genètica? Universitat de València. ISBN: 84-370-4157-0.
- Watson, J.D., Baker, T.A., Bell, S.P., Gann, A., Levine, M. y Losick, R. (2006). Biología Molecular del Gen. 5ª edición. Ed. Médica Panamericana. ISBN: 84-7903-505-6.
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- Ménsua, J.L. (2003). Genética. Problemas y ejercicios resueltos. Ed. Pearson Prentice Hall. ISBN: 84-205-3341-6.

### Additional

- Atherly, A.G., Girton, J.R. y McDonald, J.F. (1999). The Science of Genetics. Saunders College Publ.
- Dieffenbach, C. L. and Dveksler, G. S. (2003) PCR primer. A laboratory manual. Cold Spring Harbor Lab. Press.
- Gardner, E.J, Simmons, M.J. y Snustad, D.P. (2000). Principios de Genética, Alamex.
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- Hartwell, L., Hood, L., Goldberg, M.L., Reynolds, A.E., Silver, L.M. y Veres, R.C. (2008). Genetics: from genes to genomes. Ed. McGraw-Hill.
- Hawley, R.S. (2003). Advanced genetic analysis: finding meaning in a genome.
- Jorde, L.B., Carey, J.C. y Banshad, M.J. (2001) Genética Médica, 4ª ed., Editorial Elsevier España.
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- Lorente, J.A. (2004). Un detective llamado ADN. Ediciones Temas de Hoy.
- Nuez, F. i J.M. Carrillo (eds.). (2000) Los marcadores genéticos en la mejora vegetal. Ed. Universidad Politécnica de Valencia.
- Snustad, D.P., y Simmons, M.J. (2000). Principles of Genetics. 2nd edition. John Wiley & Sons, Inc.
- STRACHAN and READ. Human Molecular Genetics. 2004 (3ª ed); 2010 (4ª ed.) Garland Science/Taylor & Francis Group. La tercera edición tiene traducción al castellano (2006, Mc.Graw-Hill



Interamericana).

Weir, B. S. (1996) Genetic Data Analysis II. Sinauer Assoc.

Recursos informàtics:

1. Sociedad Española de Genética, <http://www.segenetica.es/>  
Se recomienda visitar el apartado de docencia: hay lecciones, problemas y recursos multimedia.
2. Página web del libro Genética, un enfoque conceptual. En inglés.  
<http://www.whfreeman.com/pierce3e/>  
Se encuentran recursos complementarios a los del libro, tales como animaciones, resolución de problemas y enlaces de interés.
3. DNAi.org (DNA interactive) En inglés, <http://www.dnai.org/index.htm>
4. DNA from the beginning. En ingles, <http://www.dnafb.org/>
5. Libro de texto de Genética disponible como libro electrónico en la Biblioteca de Ciencias: Griffiths, A.J.F., Wessler, S.R., Lewontin, R.C. y Carroll, S.B. (2013). Genética, 9a edición. McGraw-Hill-Interamericana. ISBN: 978-84-481-9090-3.

## ADDENDUM COVID-19

**This addendum will only be activated if the health situation requires so and with the prior agreement of the Governing Council**

### **1 y 2) Contenidos y Volumen de trabajo.**

Es mantenen els continguts inicialment arreplegats a la guia docent, excepte en pràctiques.

Si la docència fora presencial. En aquest cas, amb el propòsit de respectar l'aforament al laboratori, els grups serien dividits, i no donaríem la pràctica de laboratori número 3. Com que només s'ha eliminat una de les quatre pràctiques programades, no es canvia el pes que tenen les pràctiques de laboratori en la nota final.

Si la docència no fora presencial es donarien tots els continguts de pràctiques per videoconferència.

Es manté el pes de les diferents activitats que sumen les hores de dedicació en crèdits ECTS marcades en la guia docent original

### **3) Metodología.**

En principi les classes de problemes i de tutories de grup es faran de manera presencial. En el cas de confinament i que no es pogueren donar les classes de manera presencial, les tutories de grup i la part teòrica de les classes presencials s'impartirien per videoconferència síncrona BBC.

El test avaluable de les tutories de grup (avaluació continua) es faria fent servir l'aplicació Qüestionari de l'Aula Virtual, amb el temps acotat tal i com es s'hauria fet de manera presencial.



La resolució presencial de problemes es substituiria per l'enviament en Aula Virtual, per part del professor, de documents amb el problemes que s'haurien fet en classe, resolts pas a pas amb explicacions exhaustives. Els dubtes que pogueren sorgir es resoldrien per BBC en hores de classe o bé per correu electrònic a mena de tutoria individual.

En el cas de confinament i per tant, de una docència no presencial, les quatre pràctiques es donarien en quatre sessions per videoconferència, amb el suport de presentacions powerpoint i vídeos del web.

#### **4) Evaluación.**

L'avaluació de l'assignatura es farà tal i com estava prevista. El pes de les diferents parts de l'assignatura no es veurà modificat.

L'avaluació de les pràctiques de laboratori es farà tal i com estava inicialment prevista de les pràctiques realitzades (quatre si docència no presencial; tres si docència presencial).

L'avaluació de la part de problemes i tutories es farà de manera presencial, si l'evolució de la pandèmia ho permet.

L'avaluació continua de les tutories es farà preferentment presencialment. En el cas que no es permetira la presencialitat, es faria mitjançant l'aplicació Qüestionari de l'Aula Virtual.

En el supòsit que no es poguera fer l'examen final de manera presencial, llavors es faria com a prova escrita oberta, però distribuïda en aula virtual fent servir l'aplicació Tasca. Per a tal fi, es distribuïria de tres a quatre problemes DIFERENTS a cada estudiant per a que els resolguera en un temps acotat. La resolució hauria d'incloure una explicació detallada, en cada pas, del perquè es fa el que es fa.

Si per causes tècniques, degudament justificades, algun estudiant no poguera realitzar l'examen final, s'estudiaria la possibilitat de fer una prova alternativa que, en tot cas, seria de tipus ORAL.