1. Basic description

**Name of the course:** Genetics and Genomics  
**Module:** Life Science  
**Academic year:** 2016-2017  
**Year:** 2017  
**Term:** Third  
**Degree / Course:** Bioinformatics  
**Code:** 51308  
**Number of credits:** 6  
**Total number of hours committed:** Complete  
**Teaching language:** English  
**Lecturer:** Marta Pascual is the subject coordinator. Ferran Casals, Hafid Laayouni and Marta Pascual are participating in the theoretical and problem classes. Marta Riutort, Clara Serra and Ivon Cusco are teaching the practical classes.  
**Timetable:** Complete

2. Presentation of the course

GG is a mandatory subject of the Curriculum of the Bioinformatics grade. It will be given in the 3rd trimester of the 1st year and will include 6 credits ECTS, 3 of them of theory and 3 problems/practices.

**Objectives**

This course covers the basic principles of genetics and genomics. The teaching project of this subject aims, among other goals, to:  
Introduce the student to the main principles of Genetics  
Understand the different models of genetic inheritance.
Help the student to understand the principles of linkage and chromosome mapping.
Teach the mechanisms of chromosome and genome variation.
Provide students with information on gene interactions, inheritance of complex traits and association mapping.
This introductory course will provide students with the basis to understand more specialized courses of bioinformatics curricula.

3. Competences to be worked in the course

<table>
<thead>
<tr>
<th>General competences</th>
<th>Specific competences</th>
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<tbody>
<tr>
<td>CB1, CB2, CB4, CG1</td>
<td>CE1, CE2, CE4, CE7</td>
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</table>

I. General competences

CB1. That the students have demonstrated to have acquired the knowledge and understanding in a field of study that starts from the basis of general secondary education, and is typically at a level that although it is supported by advanced textbooks, includes some aspects that involve knowledge of the forefront of their field of study.

CB2. That the students know how to apply their knowledge to their work or vocation in a professional manner and have competencies typically demonstrated through devising and defending arguments and solving problems within their field of study.

CB4. That the students can convey information, ideas, problems and solutions to both specialist and non-specialist audiences.

CG1. That the students will acquire an intra- and interdisciplinary training in both computational and scientific subjects with a solid basic training in biology.

II. Specific competences

CE1. To acquire genetic knowledge from the gene to the organismal level, with an interdisciplinary vision and special emphasis on biomedical applications.

CE2. To manage and exploit all kinds of biological and biomedical information to transform it into knowledge.

CE4. To integrate genetics and omics data for a greater understanding of biological phenomena.
CE7. To demonstrate knowledge, skills and appropriate practices in the area of the biology of organisms and biosystems.

Learning outcomes

RA1.1, RA1.3
RA2.1, RA2.2
RA4.1

RA1.1. Validate appropriate knowledge and skills in the area of biological science.

RA1.3 Understand the stages of gene expression: phenomena of cell division and death in unicellular and multicellular organisms, regulation and use of RNA as a functional molecule.

RA2.1. Visualize, manipulate and extract biological data.

RA2.2 Improve understanding of disease onset and progression.

RA4.1. Process, manage and interpret basic omics data (genomics, proteomics, transcriptomics).

4. Contents

• Basic description of contents outlined for the curriculum

This course covers the basic principles of genetic analysis and heredity. Subjects include the interpretation of progeny ratios resulting from different inheritance patterns, the analysis of linkage and chromosome mapping, the origin and effect of chromosome and genome variation, and the understanding of the genetic complexity and the need to integrate epigenetics and gene interactions information, among others, into genetic analyses.

• Provide more detail and expand upon the description of contents

THEORETICAL CLASSES (30h)

PART I. TRANSMISSION GENETICS

Session 1. Introduction to Genetic Analysis (2h)

**Session 2. Monogenic inheritance (2h)**

**Session 3. Polygenic inheritance: independent Transmission (2h)**

**Session 4. Sex linkage (2h)**

**Session 5. Other inheritance models (2h)**

**PART II. GENETIC LINKAGE AND CHROMOSOME MAPPING**

**Session 6. Crossover and recombination (2h)**

**Session 7. Genetic maps (2h)**

**Session 8. Physical maps (2h)**

**PART III. CHROMOSOME AND GENOME VARIATION**

**Session 9. Variation in chromosome structure (2h)**

**Session 10. Variation in chromosome number (2h)**
Aneuploidy. Polyploidy: autopolyploids and allopolyploids. Applications to breeding. Rounds of genome duplication in the origin of some groups (2R hypothesis).

**Session 11. Structure of the genome (2h)**
Whole-genome sequencing. Structure of prokaryotic and eukaryotic genomes. Distribution of genes and other DNA sequences in the genome. C-value paradox. Repetitive DNA.

**Session 12. The omics revolution (2h)**

**PART IV GENE INTERACTION**

**Session 13. Gene interaction and gene networks (2h)**

**Session 14. Inheritance of complex traits (2h)**

**Session 15. Heritability and association mapping (2h)**
PROBLEMS AND SEMINARS (15h)

Discussion of questions, problems, activities and seminars, with emphasis on data analysis and interpretation, to give students the opportunity to apply the knowledge from the theoretical sessions.

PRACTICAL WORK (18h)

PR1- Mendelian genetics and linkage association studies simulating Drosophila crosses (Classical Genetics Simulator) (computer room) (3h). By using a program that simulates crosses, the students will learn:
- to program crosses to find the hereditary bases of two characters in an organism,
- to test their hypothesis and interpret results from the crosses,
- to express scientifically the results obtained and the conclusions reached.

PR2- Genomics in silico (3h). In this practice the students will learn:
- how the information on genes, their structure and function is stored in genetic databases
- how they can find this information an some applications.

PR3- Detection of structural variants (microscope lab) (3h). In this practice students will focus on the analysis of different structural alterations, using different techniques: from conventional karyotype to high density arrays. Specifically they will work on:
- Numerical aberrations using standard karyotypes
- Deletions and duplications using locus specific probes in FISH analysis
- Small CNVs (gain and losses) by different types of arrays (oligonucleotides and SNP arrays)
- Uniparental disomies and mosaicism using SNP array

PR4- Detection of point mutations (computer room and laboratory) (3h). Students will deal with exome sequencing data to unravel the genetic alteration causing a Mendelian phenotype in a given pedigree. They will discuss and use different filtering according to the Mendelian pattern of inheritance. After the identification of the variant, they will design and prepare a PCR to validate the alteration in the family.

PR5- Detection of point mutations (laboratory) (3h). This practice is related to the previous one; they will go on with the validation of the alteration found in the family in PRA5. They will use direct methods (RFLP and Sanger
sequencing) to validate the mutation and perform segregation analysis in the given pedigree.

PRA6- Students presentations (3h). The students by pairs will have worked with the program CGS to determine the hereditary bases of two characters, in this class they will explain the crosses performed and the conclusions reached using scientific language and a scientific communication structure.

5. Assessment

A series of exams are used to measure the success in meeting the course learning objectives. In order to successfully complete this course, the student must pass at least with 50 % on the final mandatory examination. All exams are compulsory.

The course assessment will be performed as follows: from 10 points, 7 points will correspond to the evaluation of the theoretical contents (6 points theoretical final exam, 1 point mid-term theoretical exam), 3 points to the evaluation of practical contents.

1 point, Evaluation of mid-term theoretical exam

6 points, Evaluation of theoretical and seminars final exam will consist in:
   a) Multiple Choice: 50%
   b) Short Questions: 50%

3 points, Evaluation of practical contents will consist in:
   Assessment: 50%
   Presentation: 50%

Students assistance to practical classes is mandatory.

Recuperation Information
Only the students that after the evaluation have not passed the course can retake the final theoretical exam in July.
## Assessment elements

<table>
<thead>
<tr>
<th>Assessment elements</th>
<th>Time period</th>
<th>Type of assessment</th>
<th>Assessment agent</th>
<th>Type of activity</th>
<th>Grouping</th>
<th>Weight (%)</th>
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<tbody>
<tr>
<td>Theoretical mid-term exam</td>
<td></td>
<td>Comp</td>
<td>Opt</td>
<td>Lecturer</td>
<td>Self-assess</td>
<td>Co-assess</td>
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<tr>
<td>Theoretical final exam</td>
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<tr>
<td>Practical work</td>
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<td>Students practical presentation</td>
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### Working competences and assessment of learning outcomes:

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<th>Learning outcomes</th>
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### 6. Bibliography and teaching resources

- Basic bibliography

- Supplementary bibliography


- Teaching resources

### 7. Methodology

Theoretical classes, seminars and practicals: Face-to-face
8. Scheduling activities

Each student will receive 63 hours of class:
30 h theoretical
15 h seminars (2 groups, 20 alumni/group, 1 teacher/group)
18 h practical lessons (2 groups, 18-20 alumni/group, 1 teacher/group)

1) Scheduling activities under the curriculum, from:
   • In the classroom: 1) Lecture classes, 2) Seminars, 3) Face-to-face tutorials, 4) “Regulated” practical classes (lab…)
   • Outside the classroom: 5) Group work, 6) Individual work (reports, exercises…), 7) Internships (outside companies), 8) Independent study
<table>
<thead>
<tr>
<th>Week</th>
<th>Activity in the classroom (Grouping/type of activity)</th>
<th>Activity outside the classroom (Grouping/type of activity)</th>
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<tbody>
<tr>
<td>Week 1</td>
<td>T: 4 hours   SEM (101) 1h 30  SEM (102) 1h 30</td>
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<td>Week 2</td>
<td>T: 2 hours   SEM (101) 1h 30  SEM (102) 1h 30</td>
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<tr>
<td>Week 3</td>
<td>T: 4 hours   SEM (101) 1h 30  SEM (102) 1h 30</td>
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<tr>
<td>Week 4</td>
<td>T: 2 hours   SEM (101) 1h 30  SEM (102) 1h 30   PR (111) 3 hours PR (112) 3 hours</td>
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<tr>
<td>Week 5</td>
<td>T: 2 hours   SEM (101) 1h 30  SEM (102) 1h 30   PR (111) 3 hours PR (112) 3 hours</td>
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<tr>
<td>Week 6</td>
<td>T: 4 hours   SEM (101) 1h 30  SEM (102) 1h 30   PR (111) 3 hours PR (112) 3 hours</td>
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<tr>
<td>Week 7</td>
<td>T: 4 hours   SEM (101) 1h 30  SEM (102) 1h 30   PR (111) 3 hours PR (112) 3 hours</td>
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<td>Week 8</td>
<td>T: 4 hours   SEM (101) 1h 30  SEM (102) 1h 30   PR (111) 6 hours PR (112) 6 hours</td>
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<td>Week 9</td>
<td>T: 2 hours   SEM (101) 1h 30  SEM (102) 1h 30</td>
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<td>Week 10</td>
<td>T: 2 hours   SEM (101) 1h 30  SEM (102) 1h 30</td>
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<tr>
<td>Week final exams</td>
<td>22/6 10-13h</td>
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