ESCI upf.
School of International Studies

TEACHING PLAN FOR

Genetics and Genomics

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

General competences	**Specific competences**
CB1, CB2, CB4, CG1	CE1, CE2, CE4, CE7

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)

Concept of genetics and genetic analysis. Heredity and variability. Classification of mutations: germ and somatic mutation. Concepts of gene and polymorphism. Model organisms. Mating systems: selfing and cross-fertilization. The cell cycle. Sexual reproduction and meiosis. Genetic importance of meiosis: generation of genetic variability.

Session 2. Monogenic inheritance (2h)
Mendel's law of segregation. Reciprocal crosses. Dominance. Genotypic and phenotypic proportions of monohybrid crosses. Backcross and testcross. Segregation studies. Probability and genetics. Pedigree analysis. Multiple alleles and their relations. Lethal alleles. Inbred lines.

Session 3. Polygenic inheritance: independent Transmission (2h)
Mendel's law of independent transmission. Genotypic and phenotypic proportions of dihybrid crosses. Chromosomal basis of independent segregation. Polyhybrid crosses.

Session 4. Sex linkage (2h)
Sex determination. Heterochromosomes. X-linkage. Y-linkage. Pseudoautosomal genes. Sex limited and sex influenced inheritance. Dosage compensation in Drosophila and mammals.

Session 5. Other inheritance models (2h)
Extranuclear genes: chloroplasts and mitochondria. Cytoplasmic mutations in human. Maternal effect. Epigenetic inheritance: maternal and paternal imprinting.

PART II. GENETIC LINKAGE AND CHROMOSOME MAPPING

Session 6. Crossover and recombination (2h)
Linked genes. Detection of linkage in a testcross. Coupling and repulsion configuration. Crossover and recombination. Calculating the frequency of recombination. Multiple crossovers. Frequency of recombination and genetic distance.

Session 7. Genetic maps (2h)
Crossover analysis with three markers. Estimating genetic distances and gene mapping. Map function. Mapping with molecular markers. SNPs map. Linkage disequilibrium and haplotype maps. Detection of linkage in human genealogies. Genome Wide Association Studies (GWAS).

Session 8. Physical maps (2h)

Physical maps. Karyotypes. Physical location of genes by in situ hybridization. Chromosome painting. Relationship between genetic maps and physical maps. Hot spots of recombination. Using fine mapping to identify genes.

PART III. CHROMOSOME AND GENOME VARIATION

Session 9. Variation in chromosome structure (2h)
Deletions, duplications, translocations and inversions: Origin, detection and genetic effect. Gene families.

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)
Genomes in databases. Annotation to identify gene sequences. Predicting gene and protein functions. Transcriptomics. Proteomics. Metagenomics.

PART IV GENE INTERACTION

Session 13. Gene interaction and gene networks (2h)
The modular nature of the gene network. Regulatory genes and modifiers. Protein / DNA and protein / protein interactions. Pleiotropy. Penetrance and expressivity. Environmental effects. Reaction norm and developmental noise. Interaction between genes responsible for a trait. The complementation test.

Session 14. Inheritance of complex traits (2h)
Dominant and recessive epistasis. Modifications to Mendelian proportions. Complex traits. The multiple-gene hypothesis for quantitative inheritance. Measuring quantitative variation.

Session 15. Heritability and association mapping (2h)
Broad-sense heritability. Narrow-sense heritability. Artificial selection. QTLs and genetic disorders. Mapping QTL in populations with known pedigrees. Association mapping in random-mating populations.

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 conclussions 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	Time period	Type of assessment		Assessment agent			Type of activity	Grouping		Weight (%)
		Comp	Opt	Lecturer	Self-assess	Co-assess		Indiv	Group (#)	
Theoretical mid-term exam										10%
Theoretical final exam										60%
Practical work										15%
Students practical presentation										15%

Working competences and assessment of learning outcomes:

																	Learning outcomes

6. Bibliography and teaching resources

- Basic bibliography

Griffiths *et al* (2015) An Introduction to Genetic Analysis. 11th ed. W.H. Freeman

- Supplementary bibliography

Vogel and Motulsky's (2010) Human Genetics, 4th edition

Klug *et al* (2016) Concepts of Genetics. 11th ed. Pearson Education

Pierce (2014) Genetics: a conceptual approach. 5th ed. W.H. Freeman

- 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

Week	Activity in the classroom Grouping/type of activity	Activity outside the classroom Grouping/type of activity
Week 1	T: 4 hours SEM (101) 1h 30 SEM (102) 1h 30	
Week 2	T: 2 hours SEM (101) 1h 30 SEM (102) 1h 30	
Week 3	T: 4 hours SEM (101) 1h 30 SEM (102) 1h 30	
Week 4	T: 2 hours SEM (101) 1h 30 SEM (102) 1h 30 PR (111) 3 hours PR (112) 3 hours	
Week 5	T: 2 hours SEM (101) 1h 30 SEM (102) 1h 30 PR (111) 3 hours PR (112) 3 hours	
Week 6	T: 4 hours SEM (101) 1h 30 SEM (102) 1h 30 PR (111) 3 hours PR (112) 3 hours	
Week 7	T: 4 hours SEM (101) 1h 30 SEM (102) 1h 30 PR (111) 3 hours PR (112) 3 hours	
Week 8	T: 4 hours SEM (101) 1h 30 SEM (102) 1h 30 PR (111) 6 hours PR (112) 6 hours	
Week 9	T: 2 hours SEM (101) 1h 30 SEM (102) 1h 30	
Week 10	T: 2 hours SEM (101) 1h 30 SEM (102) 1h 30	
Week final exams	22/6 10-13h	