

BMI: Biomedical Informatics

Descriptive details concerning the subject:

- Name of the subject: Biomedical informatics
- Code : BMI
- Type of subject: Optional
- Credits: 5
- ECTS: 5
- Total hours: 125
- Scheduling:
- Course: 1st Course
- Period: 1st term 1st 5 weeks
- Coordination: Arcadi Navarro
- Department: Ciències Experimentals i de la Salut
- Building:
- Lecture Times: 11:00-12:00

- Teaching staff:

- Language: English
- Lecturer: Arcadi Navarro
- Department: Ciències Experimentals i de la Salut

- Language: English
- Lecturer: Francesc Calafell
- Department: Ciències Experimentals i de la Salut

- Language: English
- Lecturer: Carlos Morcillo
- Department: Ciències Experimentals i de la Salut

- Language: English
- Lecturer: Belén Lorente
- Department: Ciències Experimentals i de la Salut

Subject presentation:

This course focuses in how to user numerical and computational tools to can learn about the genetic architecture of human phenotypic differences. Obviously, the most relevant such differences are those relating to disease, such as the presence/absence of the disease, differential susceptibility to it and/or differential drug responses. An emphasis will be made on the methods used to find genes associated with complex disease, which are currently being applied to study many diseases of great relevance for public health. Particular cases will be used as illustrative examples.

The course comprise 5 ECTS credits, implying 20/25 hours of plenary lectures, 10 hours of exercises and hands-on computer classes, 13/18 hours of reading and personal study, and 2 hours performing tests

The subject is based on the understanding of key methodological concepts and tools and on the application of common software found in the genetic epidemiology and statistical genomics labs around the world.

As this is a completely incremental subject, the student is advised of the need of strong interaction with the lecturers and the need of keeping the class material up to date.

The subject focuses on practical implementation of different types of tools for simple association and linkage studies. Thus, the methods used are strongly based on the good understanding of basic principles of computer programming and statistics. The course includes magistral lectures and hands-on exercises on the use of publicly available resources and software packages.

The course will be evaluated by means of an exam, individual and based on short questions/answers, some problems and some text questions.

- Prerequisites in order to follow the itinerary:

Previous programming knowledge and notions of genomics and statistics are expected.

It is advisable -but not compulsory- to have coursed BDA and BCO.

- Competences to be achieved in the subject

Instrumental:

1. Proficient reading/writing/listening scientific English related to the subject.
2. Knowledge of office software to do quality scientific presentations and reports related to the subject.
3. Elements of Statistics: concepts of association, correlation and interaction.
4. Elements of Genomics: genes, alleles, SNPs, markers, phenotypes, expression, complex traits.

Interpersonal:

1. Group work
2. Ability to solve by yourself a given problem

Systemic:

1. Analysis and synthesis abilities
2. Ability to search for information

Specific:

1. To understand the complex nature of the factors generating phenotypes.
2. To understand the concept of heritability.
3. To become familiar with the different forms of human genome variation.
4. To understand the concept of Mendelian Disease.
5. To understand the concepts of Complex Trait and Complex Disease.
6. To understand the genealogical and correlational structure of human genome variability.
7. To master the basics of Linkage Disequilibrium measures and testing.
8. To master the concept of genotype and haplotype.
9. To understand and apply the concept of Linkage Mapping
10. To understand and apply the concept of Association Test.
11. To master different allele-disease association measures.
12. To understand and apply the concept of Transmission Disequilibrium Test.
13. To understand what interactions are and how they can be tested
14. To master and apply the concept of logistic regression.
15. Ability to use extant genetic epidemiology software.
16. To understand the concept of multiple testing problems.
17. To master different methods to correct for multiple testing.

18. To get the essentials of whole genome scan approaches.

19. To master information retrieval from disease-related public databases.

- Learning aims:

To understand and apply the computational methods currently used in Biomedicine to ascertain the genetic basis of complex disease and drug response.

- Evaluation:

General assessment criteria:

The evaluation will consist of a final exam at the end of the course, worth 80% and the evaluation of the practical exercises performed during the course.

Competence Evaluation	Attainment indicator	Assessment procedure	Scheduling
Instrumental			
1. Proficient reading/writing/listening scientific English related to the subject	Correct understanding of proposed exercises and correct final presentation	Implicit in the exam and exercises	End of term
2. Knowledge of office software to do quality scientific presentations and reports.	High quality presentation of results of exercises.	Implicit in exercises	Progressive
3. Elements of Statistics: concepts of association, correlation and interaction.	Ability to follow the proposed exercises	Implicit in the exam and exercises	Progressive
4. Elements of Genomics: genes, alleles, SNPs, phenotypes, expression, complex traits.	Ability to follow the proposed exercises	Implicit in the exam and exercises	Progressive

Interpersonal

1. Group work	Ability to do team work both in programming and in preparing final presentation	Implicit in group-based exercises.	End of term
2. Ability to solve by yourself a given problem	Correct answer of set of pen and pencil exercises and final examination	Implicit in exercises and final exam	End of term

Systemic

1. Analysis and synthesis abilities	Development of algorithms for proposed problems in molecular simulations	Implicit in exercises	Progressive
2. Ability to search for information	Complete final presentation	Implicit in exercises	Progressive

Specific

1. To understand the complex nature of the factors generating phenotypes.		Exam	End of term
2. To understand the concept of heritability.		Exam	End of term
3. To become familiar with the different forms of human genome variation.		Exam	End of term
4. To understand the concept of Mendelian Disease.		Exam	End of term
5. To understand the concepts of Complex Trait and Complex Disease.		Exam	End of term
6. To understand		Exam	End of term

the genealogical and correlational structure of human genome variability.

7. To master the basics of Linkage Disequilibrium measures and testing.

Exam

End of term

8. To master the concept of genotype and haplotype.

Exam

End of term

9. To understand and apply the concept of Linkage Mapping

Exam

End of term

10. To understand and apply the concept of Association Test.

Exam

End of term

11. To master different allele-disease association measures.

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End of term

12. To understand and apply the concept of Transmission Disequilibrium Test.

Exam

End of term

13. To understand what interactions are and how they can be tested

Exam

End of term

14. To master and apply the concept of logistic regression.

Exam

End of term

15. Ability to use extant genetic epidemiology software.

Exam

End of term

16. To understand the concept of multiple testing problems.

Exam

End of term

17. To master

Exam

End of term

different methods to correct for multiple testing.

18. To get the essentials of whole genome scan approaches.

Exam

End of term

19. To master information retrieval from disease-related public databases.

Exam

End of term

Contents

Contents 1: Block 1: Overview of Health, Disease and Genomics.

Concepts

Procedures

Attitudes

Health and disease.

Diseases as phenotypes.

Disease types: the

Mendelian to complex spectrum. The common variant/ common disease paradigm.

To be able to obtain information from OMIM.

To be able to obtain information from GAD.

Quantitative trait loci (QTLs). Heritability concept.

To be able to use the basic concepts of heritability to perform basic calculations of the contributions of genetic and environmental factors to a given trait.

Variation in the human genome. Types of variation. Dynamics of variation. Sequencing and Genotyping techniques.

Linkage disequilibrium: the genealogical basis of association between Disease Susceptibility Loci and Genetic Markers

To be able to compute linkage disequilibrium between markers.

Contents 2: Block 2: Family-based methods for disease mapping.

Concepts

Procedures

Attitudes

Family studies, adoption studies, twins.	To be familiar with family- based inference.
Linkage analysis. Definition. Types. Properties.	To be able to perform basic linkage-mapping calculations.
Examples	To be able to perform basic TDTs.
The Transmission- Disequilibrium test.	

Contents 3: Block 3: New methods and tools for disease gene mapping.

Conceptes	Procedures	Attitudes
Association studies. From single polymorphisms in candidate genes to Whole Genome Scans.	To be able to infer haplotypes from genotypes. To be able to perform different association tests.	
Population genomics. The P3G projects, WTCCC, Biobanking and other public initiatives.	To be able to select candidate genes for a given disease. To master information retrieval from disease-related public sources.	
Copy number variation and disease.		

Contents 4: Block 4: Technical and computational challenges.

Conceptes	Procedures	Attitudes
Shortcomings and challenges of large- scale association studies. Replications. Interactions. Multiple testing. Functional interpretation of results. Expression studies and genetic disease	To be able to understand and implement different strategies for multiple test correction. To understand the concept of interaction and to be able to deploy different strategies to detect interactions. To be able to develop functional interpretations of gene mapping studies and to	

suggest further research avenues.

Being able to get a synthesis of the different methodologies for association studies in a critical way

Contents 5: Block 5: Examples of the Genomics of complex disease.

Conceptes

Examples on the genetics of normal variation: skin color, eye color, personality traits.

Recent studies on the genetics of disease. Pharmacogenetics.

Procedures

To be able to critically assess extant research on the genetic architecture of complex disease.

To be able to pinpoint specific drawbacks related to different computational methodologies.

Attitudes