

BASIC DETAILS:

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| Subject: | GENÓMICA | | |
| Id.: | 33696 | | |
| Programme: | DOBLE GRADO EN FARMACIA Y BIOINFORMÁTICA. PLAN 2019 | | |
| Module: | CIENCIAS DE LA VIDA | | |
| Subject type: | OBLIGATORIA | | |
| Year: | 2 | Teaching period: | Segundo Cuatrimestre |
| Credits: | 6 | Total hours: | 150 |
| Classroom activities: | 66 | Individual study: | 84 |
| Main teaching language: | Inglés | Secondary teaching language: | Castellano |
| Lecturer: | NAVARRO VERA, ISABEL (T) | Email: | inavarro@usj.es |

PRESENTATION:

Genomics is the science that studies the structure, content and evolution of genomes.

The human genome decodification represented a historic landmark which opened the doors to studies in comparative genomics, evolution of humans, genotype-phenotype association studies and for the discovery of genes or genetic regions, their functions and relation to illnesses or risks. The development of genome sequencing techniques has led to an acceleration of the knowledge in this field. Moreover, the appearance of new generation sequencing methods, allowed the production of a huge amount of genetic data and the need of bioinformatic tools to analyze and interpret them.

The main objectives of this course are to understand the diversity and complexity of genomes, to broaden the vision of genomics and the molecular and bioinformatic techniques used as well as their current applications, to show the potential applications of genomics, transcriptomics and proteomics data, and to learn the experimental and computational methods used in the so-called "omics" sciences.

PROFESSIONAL COMPETENCES ACQUIRED IN THE SUBJECT:

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| General programme competences | G03 | Cooperate to achieve common results through teamwork in a context of integration, collaboration and empowerment of critical discussion. |
| | G04 | Reason critically based on information, data and lines of action and their application on relevant issues of a social, scientific or ethical nature. |
| | G05 | Communicate professional topics in Spanish and / or English both orally and in writing. |
| | G07 | Choose between different complex models of knowledge to solve problems. |
| | G08 | Recognise the role of the scientific method in the generation of knowledge and its applicability to a professional environment. |
| | G10 | Apply creativity, independence of thought, self-criticism and autonomy in the professional practice. |
| Specific programme competences | E12 | Apply the principles and techniques of protein computational modelling to predict their biological function, their activity or new therapeutic targets (Structural Bioinformatics, Computational Toxicology). |
| | E13 | Apply omics technologies for the extraction of statistically significant information and for the creation of relational databases of biodata that can be updated and publicly accessible to the scientific community. |
| | E15 | Infer the evolutionary history of genes and proteins through the creation and interpretation of phylogenetic trees. |
| | E16 | Plan linkage and association studies for medical and environmental purposes. |
| | E17 | Induce complex relationships between samples by applying statistical and classification techniques. |
| | E18 | Apply statistical and computational methods to solve problems in the fields of molecular biology, genomics, medical research and population genetics. |
| | E19 | Explain the main biochemical reactions by applying the principles of chemical kinetics and thermodynamics. |
| | E20 | Relate the overall functioning of the organism with the basic mechanisms at the cellular and molecular level. |
| | E21 | Apply computational and data processing techniques for the integration of physical, chemical and biological concepts and data for the description and/ or prediction of the activity of a substance in a given context. |

PRE-REQUISITES:

To get a good use of the subject and to achieve a good progress, students must have basic knowledge of molecular biology and genetic.

Good skills in English to allow the understanding of the explained subject as well as the active participation in the activities, classes and works raised in the subject, are necessary

SUBJECT PROGRAMME:

Observations:

The following programm shows the differetn units in which the subject is divided. Along all of them, there will be practical lessons to show several bioinformatic applications used in the daily genomic laboratory workflow showing the students how important the bioinformatic tools are nowadays for different medical disciplines.

Due to the pandemic situation tuesday's lessons will be online.

Subject contents:

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| 1 - Introduction to Genomics |
| 1.1 - The brief history of genomics:Genomics: From The Human Genome Proyect towards 1.000,000 Genomes Proyect. |
| 1.2 - Genetics vs Genomics. |
| 1.3 - Basic concepst on Genomics. Structure. Coding and no condng regions |
| 1.4 - The viral genome. The procariotic genome. |
| 1.5 - The eucariotic genome |
| 1.6 - DNA Sequencing technology. First steps. Sanger sequencing. |
| 1.7 - NGS: sequence by synthesis and pyrosequencing. The hatching of genomics. |
| 2 - Human Genomics |
| 2.1 - Human genome structure. |
| 2.2 - Genetic variants: Types and transmission. Consequences. |
| 2.3 - How to study the human genome. Different approaches: from a single mutation to WGS. |
| 2.4 - Bioinformatics: analytical procedure-> sequencing, assembling, variant anotation and clinical interpretation . Sources, softwares, DDBB |
| 2.5 - Clinical cases: How to choose the best way. Gene, panel, CES, WES or WGS. Familial approach. |
| 2.6 - Germinal vs somatic studies. Genomics and monitorization: cancer and organ transplantation |
| 2.7 - Pharmacogenetics and pharmacogenomics |
| 2.8 - Informed consent. Genetic counselling. Ethical issues. |
| 3 - Metagenomics |
| 3.1 - Concept of species. Identifying different species. |
| 3.2 - Metagenomics and agrifood industry. |
| 3.3 - Metagenomics and healthcare. |
| 3.4 - Metagenomics and forensics. |
| 4 - Other "omic" sciences |
| 4.1 - Transcriptomics. |
| 4.2 - Proteomics. |
| 4.3 - Methylomics. |
| 4.4 - Metabolomics. |

Subject planning could be modified due unforeseen circumstances (group performance, availability of resources, changes to academic calendar etc.) and should not, therefore, be considered to be definitive.

TEACHING AND LEARNING METHODOLOGIES AND ACTIVITIES:

Teaching and learning methodologies and activities applied:

Theoretical sessions: Transmission of content through oral presentation and ICT support. Each topic will be presented in a synthetic way with a practical and scientific approach. Through practical workshops it is intended that the student relate the knowledge learned with situations related to professional life. During the sessions the teacher may require the participation of the students and the delivery of written material. The students may expose their doubts or difficulties during their development.

Tutoring sessions of optional assistance: in these sessions the student may ask the teacher in person as well as through the virtual platform questions that have not been clarified in the classroom. During this time the student may request a specific extension bibliography on a specific topic and or any other information related to the subject.

Practice sessions that will have the objective of showing geneticist activities in the laboratory at a practical level. How to use the bioinformatics tools for the analysis of genomic data and how to treat the findings as professional and for patients or specialists from different professional fields.

Student work load:

| Teaching mode | Teaching methods | Estimated hours |
|-----------------------------------|---|-----------------|
| Classroom activities | Master classes | 22 |
| | Other theory activities | 8 |
| | Practical exercises | 4 |
| | Practical work, exercises, problem-solving etc. | 6 |
| | Debates | 2 |
| | Coursework presentations | 6 |
| | Films, videos, documentaries etc. | 2 |
| | Laboratory practice | 3 |
| | Participation in seminars, conferences etc. | 2 |
| | Other practical activities | 2 |
| | Assessment activities | 4 |
| Individual study | Extra-curricular activities (visits, conferences, etc.) | 5 |
| | Tutorials | 2 |
| | Individual study | 32 |
| | Individual coursework preparation | 10 |
| | Group coursework preparation | 4 |
| | Research work | 12 |
| | Compulsory reading | 10 |
| | Recommended reading | 6 |
| | Extra-curricular activities (visits, conferences, etc.) | 4 |
| Other individual study activities | 4 | |
| Total hours: | | 150 |

ASSESSMENT SCHEME:

Calculation of final mark:

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|------------------------|--------------|
| Individual coursework: | 30 % |
| Group coursework: | 15 % |
| Final exam: | 25 % |
| Test a través de PDU: | 10 % |
| Midterm exam: | 20 % |
| TOTAL | 100 % |

*Las observaciones específicas sobre el sistema de evaluación serán comunicadas por escrito a los

alumnos al inicio de la materia.

BIBLIOGRAPHY AND DOCUMENTATION:

Basic bibliography:

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| Brown T. A. Genomes 3, 3rd edition. Oxford. .(2007) |
| Gregory T. R. The evolution of the genome. Elsevier.(2006). |
| Hartwell L., Fischer J., Aquadro C., Goldberg M. y Hood L. Genetics: from genes to genomes, 5th edition. McGraw-Hill.(2014). |
| Lesk A. M. Introduction to Genomics. Oxford University Press.(2007). |
| Klug W. S., Cummings M. R., Spencer C. A. y Palladino M. A. Concepts of Genetics, 11th edition Pearson Education, Inc. (10th edición traducida al castellano).(2014). |

Recommended bibliography:

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| Ginsburg G y Willard H. Genomic and personalized medicine, 2nd Edition. Elsevier.(2013). |
| Pierce B. A. Genetics: A conceptual approach, 5th edition. W. H. Freeman and Co. (3ª edición traducida al castellano en Editorial Médica Panamericana).(2014). |

Recommended websites:

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| PubMed | https://www.ncbi.nlm.nih.gov/pubmed |
| Genetics home reference | https://ghr.nlm.nih.gov/ |
| Pharmgkb | https://www.pharmgkb.org/ |
| Ensembl | http://www.ensembl.org/ |
| Leiden Open Variation Database | https://www.lovd.nl/ |
| Human Gene Mutation Database | http://www.hgmd.cf.ac.uk/ac/index.php |
| ClinVar | https://www.ncbi.nlm.nih.gov/clinvar/ |
| European Society of Human Genetics | eshg.org |
| American College of Medical Genetics and Genomics | https://www.acmg.net/ |