

### **IDENTIFICATION DETAILS**

Degree:	Biotechnology			
Scope	Biology and Genetics			
Faculty/School:	Experimental Sciences			
Course:	GENOMICS AND PROTEOMICS			
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Туре:	Compulsory		ECTS credits:	6
Year:	4	ſ	Code:	2039
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Teaching period:	Seventh semester			
Subject:	Advanced Biotechnology Training Technologies			
Module:	Biotechnology Tools			
Teaching type:	Classroom-based			
Language:	Spanish			
Total number of student study hours:	150			

### SUBJECT DESCRIPTION

In general, it is intended that students achieve with this subject the acquisition of the basic concepts that underlie modern Genomics and Proteomics. These disciplines aim at the global study of the Genome and the Proteome. The Genome is defined as the genetic information common to all cells of the body, and the Proteome, which constitutes the expression of the Genome, as the set of proteins that express and interact with each other under given conditions and that give each cell its individual character.

This subject is part of the Biotechnological Tools and Matter Module Advanced Biotechnological Training Technologies. These new disciplines within Molecular Biology and Genetics aim at the global study of the Genome and the Proteome from different points of view: structural, functional and evolutionary. The student will acquire a vision of how the genome of different species has been sequenced, with special reference to the human genome, as well as all the technological and bioinformatic development that this has entailed. Emphasis will be placed on the social, ethical and medical consequences of the Human Genome project and its derivatives to date. In addition, an overview will be given of current genome resequencing projects and the T2T and Pangenome project for the study of variations in the genome sequence at the individual level, and the consequences of such approaches from a social and medical point of view. In addition, the student must acquire the ability and skill to interpret the results obtained by these technologies and to consider appropriate experimental strategies to solve problems related to the study of the genome from a global perspective. Finally, it is intended that students learn about the new techniques that have emerged in recent years as well as the review of the improvements applied to classic techniques in Molecular Biology.

### GOAL

The general objective of the course is the study of classical techniques and, fundamentally, of the most current techniques and procedures used in the study and research of the genome and proteome, allowing us to acquire a complete, integrated, social and current vision of the disciplines of Genomics and Proteomics; at the same time, it aims to show their applications in the biotechnology, pharmaceutical, medical and agricultural fields. At the same time, the capacity for analysis, synthesis and deduction corresponding to this subject will be promoted, which will allow, thanks to the discussion of novel scientific works and the management and search of scientific information in databases available on the network as sources of information in these disciplines, to acquire an analytical and critical capacity for new results or scientific discoveries.

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The specific aims of the subject are:

- Historical view of the technological advances that have allowed the appearance of this new part of the knowledge of Biology.

- Knowledge of the tools necessary for the collection of information (statistical methods, etc.) .
- Knowledge of the new applications that derive from technological advances based on Genomics.
- Knowledge of specific techniques and their applications.
- Information management at the computer level of current databases.

- Know the basic foundations that support reactivity protein chemistry in relation to the experimental approaches used in Proteomics.

- Understand how different types of mass spectrometers work when analyzing proteins.

- Know how peptides and proteins behave when analyzed and fragmented in a mass spectrometer.

- Learn to interpret mass spectra, including fragmentation spectra.

- Know the set of technologies and experimental strategies used for the mass analysis and quantification of proteins.

- Learn to interpret the results of mass identification and quantification from a biological, statistical and bioinformatic point.

- Know the state of the art of technology, including advances in methods for identifying post-translational modifications, interactions between proteins and other advanced techniques.

## PRIOR KNOWLEDGE

The student who accesses the subject should have good basic training in the nature and function of proteins and nucleic acids. Therefore, they should have solid knowledge in Biochemistry, Molecular Biology and Genetics. It is also very advisable for the student to have a good level of English that allows them to keep track of the specific bibliography of the subject as well as the material provided in the classes.

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## COURSE SYLLABUS

#### GENOMICS

TOPIC 1: Introduction to Genomics. Definition and general concepts. Era of the "omics". Genomics areas of study. Prokaryotic Chromosome. Eukaryotic chromosome.

THEME 2: Mapping. Genetic maps. Physical maps. Map integration.

TOPIC 3: Sequencing. Classic Sanger method. Hierarchical method and shotgun. Human Genome Project (s). New methods of mass sequencing (NGS and 3rd generation sequencing). Short reads: Pyrosequencing (Roche), by synthesis (Illumina), ligation (ABI), ion-torrent. Long reads: PacBio SMRT and ONT. Applications and challenges of mass sequencing. Workflow in mass sequencing projects. Annotated Gene and Gene Ontology. T2T and Panprojects. New models of the human genome.

TOPIC 4. Functional Genomics I (DNA). Understanding human genetic diseases. Chromoopathies and CGH. Monogenic diseases, and linkage analysis. Complex diseases, non-parametric analyses, ligament imbalance and association studies. GWAS and PRS. ENCODE project and genomic techniques used.

TOPIC 5. Functional Genomics II (RNA). Genecode Project and derivatives. Transcriptomics of non-coding and coding genes. Gene expression studies using microarrays and RNAsec.

TOPIC 6. Epigenomics. Techniques for the study of DNA methylation, histone modification, remodeling complexes and ncRNAs. Epitranscriptomics.

TOPIC 7. Single-cell and spatial sequencing: examples in transpistomics and epigenomics. Human Cell Atlas.

TOPIC 8. Metagenomics. Genomic analysis of microbial ecosystems and its applications.

TOPIC 9. Genomics applications. Paleogenomics.

TOPIC 10. Future prospects.

#### PROTEOMICS

TOPIC 1.- Introduction to Proteomics. Proteomics concept. Historical perspective. Technological perspective. Separation of proteins. Protein Analysis. Interpretation of results. (JV)

TOPIC 2.- Protein Chemistry and Proteomics. Fundamentals of the chemical reactivity of proteins. Chemical properties of amino acids. Isoelectric Point of Proteins. Isoelectric point calculation. Reactions of interest in Proteomics.

TOPIC 3.- Experimental strategies in Proteomics. SDS-PAGE monodimensional electrophoresis. 2D-IEF-SDS-PAGE two-dimensional electrophoresis. Techniques for the preparation of two-dimensional gels. SAY. Twodimensional electrophoresis data processing

TOPIC 4.- Interpretation of mass spectra. Resolution. Accuracy. Isotopic envelope. Calculation of the isotopic envelope. Deconvolution of charges.

TOPIC 5.- Foundations of mass spectrometry. Mild ionization methods. MALDI. Electrospray. Analyzers. TOF. Quadrupole. Ion trap. Linear trap. FT. Orbitrap. Coupling of ionizers and analyzers

TOPIC 6.- Identification of proteins in gels I. Gel digestion techniques. MALDI-TOF and Peptide mass fingerprinting. Search engines. Interpretation of results.

TOPIC 7.- Molecular mechanism of peptide fragmentation. Fragmentation series. Roepstorf-Fohlman nomenclature. Fragmentation rules. Interpretation of MS/MS spectra of peptides. Identification of peptides in databases based on MS/MS spectra. Search engines, scores and error rate. Characterization of post-translational modifications

TOPIC 8.- Tandem mass spectrometers (MS/MS). Triple quadrupole. Quadrupole-TOF. Ionic and linear trap. TOF-TOFF.. Types of fragmentation. CID. LIFT. ETD. Multiple fragmentation. Scan modes. Parent scan. Neutral loss scan. Identification of proteins in gels using liquid-mass chromatography and fragmentation.

TOPIC 9.- Second Generation Proteomics. European school and American school. Multidimensional chromatography. Massive proteome analysis using second generation.

TOPIC 10.- Differential expression proteomics using second-generation techniques. Stable isotopic dilution. Chemical methods: ICAT. iTRAQ. Metabolic methods: SILAC. Enzymatic methods: 18O. Determination of differential expression changes.

TOPIC 11.- Applications of proteomics in biomedicine. Practical Proteomics Classes 1.- Protein chemistry and isoelectric point (DP) 2.- Deconvolution of mass spectra (EB) 3.- Calculation of the isotopic envelope (EB) 4.- Identification of proteins using peptide mapping (PMF) (AM) 5.- Interpretation of MS/MS spectra of peptides (PM) 6.- Characterization of post-translational modifications (PM) 7.- Identification of peptides in databases by MS/MS (MT)

# **EDUCATION ACTIVITIES**

AF1- Participatory theoretical expository classes

AF2- Practical classes: exercises, case studies and/or experimental work carried out in the laboratory.

AF4- Seminars, round tables, workshops, tutorials, debates

AF5- Evaluation

AF6- Stand-alone study

"THE TEACHERS OF THE SUBJECT DO NOT AUTHORIZE THE PUBLICATION BY THE STUDENT OF THE MATERIAL PROVIDED BY THE TEACHERS OF THE SUBJECT IN THE VIRTUAL CLASSROOM, OR BY ANY OTHER MEANS"

## DISTRIBUTION OF WORK TIME

TEACHER-LED TRAINING ACTIVITIES	INDIVIDUAL WORK		
60 Horas	90 Horas		

### SKILLS

#### **Basic Skills**

Students must have demonstrated knowledge and understanding in an area of study that is founded on general secondary education. Moreover, the area of study is typically at a level that includes certain aspects implying knowledge at the forefront of its field of study, albeit supported by advanced textbooks

Students must be able to apply their knowledge to their work or vocation in a professional manner and possess skills that can typically be demonstrated by coming up with and sustaining arguments and solving problems within their field of study.

Students must have the ability to gather and interpret relevant data (usually within their field of study) in order to make judgments that include reflections on pertinent social, scientific or ethical issues

Students must be able to convey information, ideas, problems and solutions to both an expert and non-expert audience

Students must have developed the learning skills needed to undertake further study with a high degree of independence

To be familiar with the applications of biotechnology in the healthcare, food, agrobiotechnological, environmental and chemical fields.

To understand the social, economic and environmental implications of professional activity.

To understand the ethical implications of professional and personal activity.

Capacity for teamwork and group management.

To have acquired the ability for analytical, synthetic, reflective, critical, theoretical and practical thought.

Capacity for problem-solving and decision-making.

To recognize the mutual influence existing between science, society and technological development in order to strive for a sustainable future.

To develop capacity for and a commitment to learning and personal development.

To develop an ability to search for, take in, analyze, sum up and relate information.

To develop oral and written communication skills.

To be familiar with the applications of biotechnology in the healthcare, food, agrobiotechnological, environmental and chemical fields.

To understand the social, economic and environmental implications of professional activity.

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To develop capacity for and a commitment to learning and personal development.

To develop an ability to search for, take in, analyze, sum up and relate information.

To develop oral and written communication skills.

#### Specific skills

Know and understand the applicability of multidisciplinary techniques that include concepts of protein chemistry, mass spectrometry, protein treatment and manipulation, biostatistics and bioinformatics.

Understand the foundation and applications of microarrays in biotechnology.

Know and know how to apply new genomic techniques to the fields of medicine, biology, pharmacy and agriculture.

Learn about the set of technologies and experimental strategies used for the mass analysis and quantification of proteins.

Develop habits of rigorous thinking.

Ability to communicate the knowledge acquired orally and in writing.

Know how to apply the theoretical knowledge acquired to solving problems and practical cases related to different subjects.

### LEARNING RESULTS

It describes the study of the genome and the proteome, as well as their global integration into cellular processes.

Explain the multidisciplinary nature of these subjects, integrating data provided by several traditional disciplines.

Describes the most commonly used study methods currently in Genomics and Proteomics.

Discusses scientific contributions to the fields of Genomics and Proteomics.

It uses bioinformatics managing databases derived from both disciplines.

Describes the post-genomic vision (the study of cellular functioning and development of organisms based on the study of Genome and Functional Genomics).

It solves problems or practical issues related to the proteomic and genomic techniques described.

## LEARNING APPRAISAL SYSTEM

1. Ordinary evaluation system

The global evaluation system for the subject will be:

SE1- Evaluation of the theoretical content of the subject 80%;

S3- Evaluation of seminars: carrying out and presenting exercises, case studies, debates, tutorials 20% For the evaluation of the subject, the student's performance will be taken into account in all the activities proposed

throughout the course.

2. Alternative evaluation system

In the case of repeating the course, the Genomics or Proteomics grade will not be saved if you have an approved part. Students in second or subsequent enrollment must contact the teacher to request to take advantage of the alternative system, which will be defined by the teacher.

Plagiarism, as well as the use of illegal means in evaluation tests, will be sanctioned in accordance with those established in the Evaluation Regulations and the University's Coexistence Regulations.

The final grade for each part will be given by the sum weighted by percentage of the score of the theoretical exam and the grade obtained in the activities proposed by each teacher in each part of the subject. In an ordinary call, the final grade of the course will be given by the average obtained in the final exam of individual Genomics and Proteomics and will be considered the final grade for the subject (Genomics and Proteomics), provided that both parts of the course have been passed with a minimum. If the teachers of the subject consider that any of them have not been adequately exceeded with this minimum (which will be determined after an evaluation of the distribution of grades in the group), the subject will be suspended without taking the average. However, the grade of that part of the subject, Genomics or Proteomics, that has been exceeded by that minimum in the ordinary call will be saved for the extraordinary call. 2. Alternative evaluation system. In the case of repeating the course, the Genomics or Proteomics grade will not be saved if you have an approved part. Students in second or subsequent enrollment must contact the teacher to request to take advantage of the alternative system, which will be defined by the teacher.

SPELLING CORRECTION CRITERIA (PAU LOE Guidelines 2009/10): Each spelling error will subtract 0.25 points from the final grade of the exercise and the errors in the accents 0.15 points, up to a maximum of 3 points in both cases. The same repeated fault will be taken into account only once. The repetition of misspellings may even result in the qualification of suspense. Abbreviations, syntactic errors, grammatical errors... will be penalized with the subtraction of 0.15 points. Plagiarism, as well as the use of illegitimate means in evaluation tests, will be sanctioned in accordance with those established in the Evaluation Regulations and the University's Coexistence

Regulations.

# ETHICAL AND RESPONSIBLE USE OF ARTIFICIAL INTELLIGENCE

1.- The use of any Artificial Intelligence (AI) system or service shall be determined by the lecturer, and may only be used in the manner and under the conditions indicated by them. In all cases, its use must comply with the following principles:

a) The use of AI systems or services must be accompanied by critical reflection on the part of the student regarding their impact and/or limitations in the development of the assigned task or project.

b) The selection of AI systems or services must be justified, explaining their advantages over other tools or methods of obtaining information. The chosen model and the version of AI used must be described in as much detail as possible.

c) The student must appropriately cite the use of AI systems or services, specifying the parts of the work where they were used and describing the creative process followed. The use of citation formats and usage examples may be consulted on the Library website(<u>https://www.ufv.es/gestion-de-la-informacion\_biblioteca/</u>).

d) The results obtained through AI systems or services must always be verified. As the author, the student is responsible for their work and for the legitimacy of the sources used.

2.- In all cases, the use of AI systems or services must always respect the principles of responsible and ethical use upheld by the university, as outlined in the <u>Guide for the Responsible Use of Artificial Intelligence in Studies at UFV</u>. Additionally, the lecturer may request other types of individual commitments from the student when deemed necessary.

3.- Without prejudice to the above, in cases of doubt regarding the ethical and responsible use of any AI system or service, the lecturer may require an oral presentation of any assignment or partial submission. This oral evaluation shall take precedence over any other form of assessment outlined in the Teaching Guide. In this oral defense, the student must demonstrate knowledge of the subject, justify their decisions, and explain the development of their work.

## **BIBLIOGRAPHY AND OTHER RESOURCES**

#### Basic

Pevsner, Jonathan (1961-) Bioinformatics and functional genomics/4th ed. Oxford:Wiley-Blackwell, 2015. (Pevsner, Jonathan (1961-) Bioinformatics and functional genomics/4th ed. Oxford:Wiley-Blackwell, 2015. , ||Brown, Terry A. Genomes 4/4th ed. New York; London: Garland Science, 2018. )

Westermeier, Reiner. Electrophoresis in Practice: Guide to Methods and Applications of DNA and Protein Separations, A [Electronic Resource]/[S. I.] :WileyVCH Verlag GmbH & Co. KGaA, 2016.

KYTE, Jack. Structure in protein chemistry/New York; London: Garland, 1995.

Krebs, Jocelyn E. Lewins Genes XI/Burlington: Jones & Bartlett, 2014.

Lesk, Arthur M. Introduction to Genomics/Oxford: Oxford University Press, 2007. (Lesk, Arthur M. Introduction to Genomics/Oxford: Oxford University Press, 2007., ||Strachan, Tom. Human molecular genetics/5th ed. Boca Raton (Florida) :CRC, 2019.)

Sudbery, Peter. Human molecular genetics/2nd ed. Madrid:Pearson Education, 2004. (Sudbery, Peter. Human molecular genetics/2nd ed. Madrid:Pearson Education, 2004., ||Korf, Bruce R. Human genetics and genomics [Electronic Resource]/[S. I.] :Wiley-Blackwell, 2013.)

Pevsner, Jonathan (1961-) Bioinformatics and functional genomics/4th ed. Oxford:Wiley-Blackwell, 2015.

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Michael Kinter, Nicholas E. Sherman. Protein sequencing and identification using tandem mass spectrometry/Wiley.

Westermeier, Reiner. Electrophoresis in Practice: Guide to Methods and Applications of DNA and Protein Separations, A [Electronic Resource]/[S. I.] :WileyVCH Verlag GmbH & Co. KGaA, 2016.

KYTE, Jack. Structure in protein chemistry/New York; London: Garland, 1995.

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