

IDENTIFICATION DETAILS

Degree:	Pharmacy			
Scope	Pharmacy			
Faculty/School:	Experimental Sciences			
Course:	HUMAN GENETICS			
Туре:	Optional		ECTS credits:	3
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Year:	5		Code:	2566
		_		
Teaching period:	Ninth semester			
Subject:	Biologics			
Module:	Biologics			
Teaching type:	Classroom-based			
Language:	Spanish			
Total number of student study hours:	75			

SUBJECT DESCRIPTION

This course will focus on the fundamental aspects of human genetics. The principles and methods of both classical and modern genetics will be studied, with a particular emphasis on their applications in the field of human biology and medicine.

Fundamental concepts will be addressed such as genetic variation and population genetics, the inheritance patterns of monogenic and polygenic diseases, chromosomopathies, the foundations and usefulness of cytogenetic and molecular methods in the field of medical genetics... etc. During the development of theoretical classes, clinical examples will be used to illustrate the importance of the topics covered in the area of biomedicine.

The fundamental objective of the Human Genetics course will be for students to understand the need for knowledge of genetics for the advancement of biomedical sciences, considering its relationship with human phenotypic variability, health and disease.

PRIOR KNOWLEDGE

It is recommended that students who access the subject of Human Genetics have solid knowledge of Cell and Molecular Biology, and Physiology and Pathophysiology.

COURSE SYLLABUS

Topic 1. Individual genetic variation: mutation and polymorphism.

Theme 2. Genetic variation in populations. Hardy-Weinberg equilibrium, factors that modify it and applications.

Theme 3. Genetic disorders with monogenic inheritance. Atypical models of monogenic inheritance.

Topic 4. Genetic disorders with complex inheritance.

Topic 5. Clinical cytogenetics: numerical and structural anomalies. Chromoopathies.

Theme 6. Genetics and cancer.

Topic 7. Prenatal diagnosis and genetic counseling.

Topic 8. Treatment of genetic diseases. Gene therapy.

Topic 9. Genetics of some physiological processes. Globin gene expression. Genetics of pigmentation. Body weight regulation. Gene expression changes during exercise. Genetics of aging.

Practical syllabus (practical seminars):

-Study of the human karyotype.

-Analysis of family trees.

-Analysis of polymorphisms and population genetics.

EDUCATION ACTIVITIES

The classes of the Human Genetics course will make use of a combined methodology in order for students to achieve the proposed objectives. The face-to-face hours will be divided between the master classes given by the teacher of the subject, and the sessions focused on the discussion of issues (practical seminars) and the oral presentation of papers prepared by the students during non-face-to-face work time. In addition, the course's website will serve as a fundamental support tool for learning.

Theory classes (AFP1) Seminars and exhibition of works (AFP4) Tutoring (AFP5) Conducting exams (AFP6) Study of theory, exercises and problems (AFNP1) Work preparation (AFNP3) Tutoring preparation (AFNP4)

Detailed description: Theory classes: master classes given by the teacher in which the fundamental concepts of the subject will be presented. These classes will be supported by computer presentations that will be available to students through the subject's website. Classes of exercises and problems: after the presentation of the fundamental theoretical concepts, students will be proposed to discuss and solve questions, practical cases and problems related to the contents of the subject. Preparation and presentation of works: students must prepare, individually or as a group, a work on a topic of interest and present it in class. The participation of all students in the scientific discussion following the exhibition will be encouraged. Tutoring: through tutoring, the teacher, at the request of the student and at the established time for this purpose, will answer questions or discuss the questions posed to him by the student, in order to guide him in learning the subject. The tutoring schedule can be consulted in the degree coordinator and will be informed by the teacher at the beginning of the course.

DISTRIBUTION OF WORK TIME

TEACHER-LED TRAINING ACTIVITIES	INDIVIDUAL WORK
35 Horas	40 Horas

Cross Skills

To nurture an attitude of intellectual curiosity and a quest for truth in all areas of life.

To be able to approach a subject by means of rigorous, profound and comprehensive thought.

To be able to assess knowledge acquired.

LEARNING RESULTS

Develop skills to identify therapeutic targets and biotechnological drug production, as well as for the use of gene therapy.

SPECIFIC LEARNING RESULTS

Know the structure of the human genome

Understand the flow of genetic information and its regulation ||Explain the causes of interindividual genetic variability and the genetic evolution of human populations.

Recognize the general laws governing the transmission of hereditary traits as well as the different types of chromosomal anomalies and analyze their clinical importance.

Give examples of diseases with both mono and polygenic inheritance, and analyze the importance of genetic factors both in physiological processes and in the development of complex pathologies.

Learn genomic technologies applied to drug discovery and development

LEARNING APPRAISAL SYSTEM

REGULAR EVALUATION SYSTEM

It is the priority system applicable to all students and is based on continuous evaluation, distributing the final grade of the subject into several sections. An important part is the evaluation of the knowledge acquired during theoretical classes. Additionally, the preparation and oral presentation of papers, and the discussion of issues and problems in practical seminars, will be evaluated. Thus, for the calculation of the final grade of the subject, the weighting of the different sections will be as follows: -

Written exam (SE1): 65%

Performing daily activities and solving exercises in practical seminars (SE2): 5% Preparation and oral presentation of the works carried out (SE3): 25% Attendance, active and relevant participation in theoretical classes (SE4): 5%

The daily activities, exercises and work that are delivered after the deadline established for this purpose will NOT be taken into account for the evaluation. This weighting will be applicable to the ordinary call, provided that the student attends at least 80% of the classes and activities carried out in the classroom. In the event of non-attendance and/or in an extraordinary call, students should contact the teacher to find out about the evaluation criteria specific to their case. The course is approved with an overall grade equal to or greater than 5.0.

ALTERNATIVE EVALUATION SYSTEM Students in second or subsequent enrollment must contact the teacher to request to take advantage of this system. The weighting of the different sections for the calculation of the final grade of the subject will be as follows:

Written exam (SE1): 65%

Performing daily activities and solving exercises in practical seminars (SE2): 5% Preparation and oral presentation of the works carried out (SE3): 25% Tutoring attendance and active participation (SE4): 5%

IMPORTANT Plagiarism, as well as the use of illegitimate means in evaluation tests, will be sanctioned in accordance with the university's Evaluation Regulations and 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

Klug, William S. Concepts of Genetics/8th ed. Madrid [etc] : Prentice Hall, 2006.

(Klug, William S. Concepts of Genetics/8th ed. Madrid [etc] :Prentice Hall, 2006. , ||Rubén Lisker, Patricia Grether González, Alejandro Zentella Dehesa. Introduction to Human Genetics/3rd edition. Mexico: The Modern Manual, 2013.)

Peter Sudbery; translated by José Luis Paternáin. Human molecular genetics/2nd ed. Madrid: Pearson Education,

2004.