VNIVERSITAT DE VALÈNCIA

Course Guide
33065 Human genetics

COURSE DATA

Data Subject	
Code	33065
Name	Human genetics
Cycle	Grade
ECTS Credits	5.0
Academic year	2022 - 2023

Study (s)

Degree	Center	Acad. year	Period
1100 - Degree in Biology	Faculty of Biological Sciences	4	First term

Subject-matter

Degree	Subject-matter	Character
1100 - Degree in Biology	16 - Fundamentals of health biology	Optional

Coordination

Name	Department
MOLTO RUIZ, MARIA DOLORES	194 - Genetics

SUMMARY

The subject Human Genetics is taught in the fourth course of the Biology degree (Plan 2009) and belongs to the intensification Fundamentals of Health Biology (FHB). It is a theoretical and practical subject of the first semester of the course along with the subjects Reproduction and Endocrinology and Clinical Biochemistry.

Human Genetics studies heredity and variation of biological traits in humans' beings, occupying a central position in Sanitary Biology and Biomedicine. The knowledge of language and concepts of Human Genetics, as well as the incorporation of the genetic and genomic perspectives of health and disease, has lead to establish a framework for learning which is essential in the health professional practice nowadays.

Human Genetics is a very broad field of biology, where traditional methodologies and more innovative strategies coexist in perfect harmony. This has allowed the study of a large number of genetic alterations responsible for disease, enabling the development of strategies for the detection, diagnosis and treatment of such pathologies. The Human Genome Project has definitively promoted the research on the isolation of genes causing diseases, initially Mendelian-type, and, more recently, diseases with complex inheritance, much more prevalent in the human population. The nature of the continuous progress in this field has high social impact, and issues related to human genetics have a constant presence in the media

and are subject to debate, not only for the scientific community but also for public opinion.

PREVIOUS KNOWLEDGE

Relationship to other subjects of the same degree

There are no specified enrollment restrictions with other subjects of the curriculum.

Other requirements

You must have 120 ECTS approved

OUTCOMES

1100 - Degree in Biology

- Conocer y saber aplicar el método científico.
- Capacidad de organización, planificación y gestión de la información usando bases de datos bibliográficas adecuadas.
- Utilización del vocabulario específico de la Biología sanitaria.
- Capacidad de resolución de problemas y toma de decisiones.
- Capacidad de elaborar artículos, informes o proyectos y de exponerlos a diferentes auditorios.
- Habilidad para el trabajo en equipo y en contextos multidisciplinares.
- Capacidad de análisis crítico de textos científicos.
- Aprendizaje autónomo y adaptación a nuevas situaciones.
- Potenciar la creatividad, iniciativa y espíritu emprendedor.
- Apreciación del rigor, el trabajo metódico, y la solidez de los resultados.
- Potenciación de la capacidad de liderazgo.
- Capacidad de utilización de herramientas matemáticas y estadísticas.
- Reflexión ética sobre la actividad profesional.
- Conocimiento de bases de legislación relacionada con la Biología.
- Saber analizar datos usando herramientas estadísticas apropiadas.
- Conocimiento de sistemas de gestión en tareas profesionales en Biología sanitaria.
- Conocer los principales métodos y técnicas experimentales aplicadas al estudio de las enfermedades humanas, su etiología y la efectividad de los tratamientos.
- Conocimiento de las enfermedades y disfunciones más frecuentes durante las distintas etapas de la vida.

- Diferenciar entre enfermedades cromosómicas, genéticas de transmisión mendeliana y multifactoriales.
- Conocer las estrategias genéticas para la prevención de enfermedades hereditarias como fundamento del consejo y diagnóstico genético.
- Conocer los fundamentos de terapia génica.
- Conocer las aplicaciones de los análisis genéticos en la identificación de individuos y la determinación de relaciones de parentesco.

LEARNING OUTCOMES

About knowledge

- Knowing and applying correctly the vocabulary and terminology specific to Human Genetics.
- Acquisition of essential knowledge about genetic and molecular bases of inherited diseases.
- Acquisition of basic knowledge about the origin of karyotypic alterations and its application in clinical cytogenetics.
- Acquisition of knowledge and its application about the methodology developed for identification of Mendelian genes and genetic risk factors in multifactorial diseases and cancer.
- Acquisition of basic knowledge of strategies to address the prevention and treatment of genetic diseases.
- Acquisition of knowledge about the genetic basis of interindividual differences in response to different drugs and its application in the context of personalized medicine.

About procedure

- Identification of the different Mendelian inheritance patterns, as well as the factors that can alter them, based on the study of genealogies and assignment of genotypes.
- Assessment of genetic prediction and recurrence risk in hereditary diseases.
  Recognition of loss of function vs gain of function mutations in hereditary diseases.
- Mapping a disease gene from the results of the different methodologies of genetic mapping.
- Distinguishing between pathological mutations and genetic polymorphisms.
  Learning the protocol for obtaining human karyotype and chromosomal abnormalities identification.
- Learning different genotyping techniques.
- Knowing the different techniques of molecular markers analysis and their application in determining biological paternity and identification of individuals.
  Interpretation and presentation of genetic analysis results form different types of samples and tests applied in the genetic diagnosis.
- Management of bibliographic sources related to human genetics from books, journals and database.

DESCRIPTION OF CONTENTS

1. Introduction

Genetics and Health Biology. Technical and scientific advances in Human
Genetics. The Human Genome Project. Largest international projects in genomics. Genomic databases. Genetic and hereditary diseases. Disease Prevalence and Incidence.

2. Chromosomal and genomic basis of diseases: Autosomes alterations

The human chromosomes. Gametogenesis and mutagenesis. Karyotype abnormalities and study tecniques. The structural anomalies of autosomes. The numerical anomalies of autosomes. Uniparental disomy and diploidy. Major cytogenetic syndromes.

3. Chromosomal and genomic basis of diseases: Sex chromosome disorders

Dosage compensation. X-chromosome inactivation. Abnormalities in the heterochromosomes. Fragile sites. Fragile X syndrome.

4. The genetic and molecular bases of inherited diseases

Mendelian inheritance patterns. Human pedigree analysis. Genetic prediction and risk of recurrence. Loss and gain of function mutations. Variations on Mendelian inheritance. Penetrance. Expressivity. Locus heterogeneity. Genetic anticipation. Pleiotropy. Genetic imprinting. Lethality. Mosaicism. Mitochondrial inheritance.

5. Genetic diseases with complex inheritance

Continuous and discontinuous multifactorial inheritance. Genetic vulnerability factors. Familial clustering. Twin studies: concordance rates and heritability. Adoption studies. Congenital malformations. Multifactorial disorders of adult life.

6. Blood groups

AB0 and Rh blood groups. . Incompatibilities.

7. Genetic identity

The application of molecular markers for genetic fingerprinting. DNA profiling as a tool to assist in the identification of individuals. Assigning probabilities in forensic genetics. Assigning probabilities in paternity testing and biological relationship.

8. Genetics of cancer

Genetic basis of cancer. Hereditary and sporadic cancer. Environmental risk factors. Oncogenes and tumor suppressor genes mutations. Alterations in DNA repair. Chromosomal instability. Tumor progression

9. Cloning human disease genes

Identification of Mendelian diseases genes: Positional cloning. Genetic mapping: Linkage analysis. Exome/genome sequencing. Sequencing of gene panels. Identification of genetic risk factors in the multifactorial diseases: association studies. Ethical issues.

10. Prevention of genetic diseases

Genetic counseling. Determining recurrence risks. Genetic diagnosis: preimplantational, prenatal and presymptomatic. Diagnostics and laboratory tests. Genetic screening in populations.

11. Treatment of genetic diseases

Treatment strategies. Gene therapy: types. Therapeutic genes and constructs. Transfer vectors and methods. Candidate diseases. Applying genomics to individualize cancer therapy. Genomic edition. Mitochondrial replacement therapy. Pharmacogenetics and personalized medicine. Ethical issues in human genetics.

12. Laboratory sessions

LABORATORY 1. Genotyping ADH3 polymorphisms by dot blot hybridization and ASO probes.gDNA amplification by PCR. Dot blot. Hybridization using Allele Specific Oligonucleotides (ASO) probes. Hybrid detection by colorimetry. Results interpretation.
LABORATORY 2. Genetic Identity.
Genotyping of a DRD4 microsatellite. Identification of X and Y chromosomes using molecular markers. Results interpretation. Elaboration of individualized molecular profiles, based on the results of the two laboratory results, for the assignment of the identity of a reference sample.

13. Problems and case studies

Problems and case studies
Pedigree analysis and determining inheritance patterns. Genetic risk and recurrence risk assessments in Mendelian diseases and chromosomal disorders. Linkage analysis. Recombination frequency calculation. Determination of LOD values. Case-control association studies: Odd ratiocalculation. Assigning probabilities in forensic genetics and paternity testing. Indirect genetic diagnosis by using molecular markers.

WORKLOAD

ACTIVITY	Hours	% To be attended
Theory classes	27,00	100
Laboratory practices	15,00	100
Classroom practices	6,00	100
Tutorials	2,00	100
Study and independent work	35,00	0
Preparation of evaluation activities	20,00	0
Preparing lectures	8,00	0
Preparation of practical classes and problem	8,00	0
Resolution of case studies	4,00	0
TOTAL	**125,00**	

TEACHING METHODOLOGY

The following teaching methods will be used to develop this subject:

1. **Theory classes**. The teacher will present the most relevant contents for the subject, using audiovisual equipment for agile development. The material necessary for proper monitoring of the lectures will be previously available in Aula Virtual.

2. **Laboratory classes**. These classes will take place in 5 sessions of 3 hours each. Attendance is compulsory for laboratory classes.

3. **Problems classes**. In these 1-hour sessions, the concepts presented in the theory classes will be reinforced and the active participation of the students will be stimulated through the discussion and resolution of problems and clinical cases. The teacher will prepare a series of exercises for each thematic block, which will allow working individually (through student personal preparation) and collectively (through exposure and discussion in the group class).

4. **Group mentoring**. It will be revised those concepts that are more complex or more difficult for students using two sessions, one hour each distributed along the course.

5. **Personalized tutoring.** Students will be encouraged to use this resource for advice and discussion with the teacher of any topic about the program, the course, or degree studies.

EVALUATION

1. Evaluation of the knowledge of theory

The concepts learned in the theoretical sessions will be evaluated through a written test at the end of the semester. The test will consist of short answer and multiple-choice questions. The value of this test will be 60% of the final mark for the subject.

2. Evaluation of practical skills

The value of this part will be 40% of the final mark for the course.

The student's ability to solve genetic questions and problems will be evaluated through a written test at the end of the semester along with the exam of theory. The value of this test is 20% of the final course grade.

Laboratory learning will be assessed taking into account the presentation of laboratory results and their discussion, 15 days after the end of these classes (10% of final mark), and a written test to be carried out in the last session of the laboratory classes (10% of final mark). Attendance to lab sessions is a prerequisite to pass the course.

Other considerations: The final mark will be the sum of the marks achieved in the different sections. To pass the course, it will be necessary to obtain a global mark equal to or greater than 5 out of 10, as long as the mark for theoretical and practical knowledge (both problems and laboratory) is independently, equal to or greater than 4 out of 10.

For students who have not passed the subject in the first call, the marks of the knowledge of theory an practical skills will be saved for the second call if it is equal to or greater than 4 out of 10. The mark obtained from the laboratory skills is saved for the next year, if it is equal to or greater than 5 out of 10. Students, who do not assist to any part of the final exam (theory or practice) and do not pass the course, will have NOT EVALUATED in the records.

ADVANCE CALL: To request the advance call of this subject, students should have performed the mandatory activities listed in this teaching guide.

REFERENCES

Basic

- JORDE LB, CAREY JC y Bamshad MJ. Genética Médica. 2021 (6ª ed). Editorial Elsevier España SL. ISBN 978-84-9113-797-9; eISBN: 978-84-9113-880-8.

-NUSSBAUM R.L., McINNES R.R., WILLARD H.F. Genética en Medicina. 2016 (8ª Ed). Elsevier España. ISBN: 9788445826423.

-PIERCE B.A. Genetics: A Conceptual Approach. 2018 (5ª Ed.). WH Freeman. ISBN 1319187811. La cuarta edición tiene traducción al castellano: Genética: un enfoque conceptual. 2016 (4ª ed). Editorial Médica Panamericana.

-PRITCHARD D.J. / KORF B.R. Genética Médica. 2015 (3ª ed). Editorial Médica Panamericana. ISBN 9788479033958.

-STRACHAN T, GOODSHIP J, and CHINNERY P. 2015. Genetics and Genomics in Medicine. Garland Science, Taylor & Francis Group, LLC. ISBN 978-0-8153-4480-3.

-STRACHAN and READ. Human Molecular Genetics. 2018 (5ª ed, ISNB 0815345895); 2010 (4ª ed, ISBN, 9780815341499) Garland Science/Taylor & Francis Group. La tercera edición tiene traducción al castellano (2006, Mc.Graw-Hill Interamericana, ISBN970-10-5135-1).

-Peter Turnpenny and Sian Ellard. Emery: Elementos de genética médica. 2018 (Ed. 15). Elsevier. ISBN 9788491132066

Libros electrónicos:
- Relación de libros electrónicos sobre temas de biomedicina. https://www.ncbi.nlm.nih.gov/books/

Additional

- -CRESPILLO MÁRQUEZ Manuel C y BARRIO CABALLERO Pedro A. (editores). GENÉTICA FORENSE. Del laboratorio a los Tribunales. 2019. Editorial Díaz de Santos. Madrid. ISBN: 978-84-9052-213-4.
-DELGADO RUBIO A., GALÁN GÓMEZ E., GUILLÉN NAVARRO E., LAPUNZINA BADÍA PABLO D., PENCHASZADEH VICTOR B., ROMEO CASABONA CARLOS MARÍA, EMALDI CIRIÓN AITZIBER. Asesoramiento Genético en la práctica médica. 2012. Editorial Médica Panamericana.
-LORENTE, J.A. Un detective llamado ADN. 2004, Ediciones Temas de Hoy. ISBN: 84-8460-386-5.

WEBS recomendadas

GeneCards: http://www.genecards.org/
Gene Names: www.genenames.org

GeneReviews: http://www.genereviews.org/
OMIM - Online Mendelian Inheritance in Man: https://omim.org/
Orphanet (portal europeo de información de referencia en enfermedades raras y medicamentos huérfanos):
https://www.orpha.net/consor/cgi-bin/index.php?lng=ES