

**COURSE DATA****Data Subject**

Code	33065
Name	Human genetics
Cycle	Grade
ECTS Credits	5.0
Academic year	2019 - 2020

Study (s)

Degree	Center	Acad. year	Period
1100 - Graduado/a en Biología	Faculty of Biological Sciences	4	First term

Subject-matter

Degree	Subject-matter	Character
1100 - Graduado/a en Biología	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 - Graduado/a en Biología

- 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 from 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.. 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. 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: molecular aspects. 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

IAB0 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: 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. Mitochondrial replacement therapy. Pharmacogenetics and personalized medicine. Ethical issues in human genetics.

12. Laboratory sessions

LABORATORY 1. Genotyping ADH3 polymorphisms by filter hybridization and ASO probes. Genomic DNA isolation from buccal mucosa cells. DNA amplification by PCR. Dot blot. Hybridization using ASO probes. Hybrids detection by colorimetry. Results interpretation.

LABORATORY 2. Genetic Identity.

Genotyping of a DRD4 microsatellite. Identification of X and Y chromosomes using molecular markers. Blood group determination by immunological techniques. Results interpretation and obtaining individualized molecular profiles 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: Odds ratio calculation. Assigning probabilities in forensic genetics and paternity testing. Indirect genetic diagnosis by using molecular markers.

WORKLOAD

ACTIVITY	Hours	% To be attended
Theory classes	26,00	100
Laboratory practices	22,00	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).

3. 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.

4. 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 studied in the theoretical sessions will be evaluated at the end of the semester by a written test. The value of this test is **60%** of the final grade for the course.

2. Evaluation of practical skills

2.1. 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.

2.2. Laboratory learning will be assessed taking into account the presentation of practical results and discussion (**10%** of final mark) and a written test (**10%** of final grade) to be held in conjunction with the previous examinations. The value of the practical skills is **40%** of the final grade for the course.

Attendance to lab sessions is a prerequisite to pass the course.

Other considerations: The final grade is the sum of the grades achieved in the different sections. To pass the course is necessary to obtain an overall rating equal to or greater than 5 out of 10, taking into account that the note of the skills and knowledge are either, independently, 4 or more out of 10, as reflected in the table. For students who have not passed the subject in the first call, the notes of the knowledge of theory and practical skills will be saved for the second call if the mark is equal to or greater than 4 out of 10. The grade 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*. 2016 (5ª ed). Editorial Elsevier España SL. ISBN 9788491130581.
- NUSSBAUM R.L., McINNES R.R., WILLARD H.F. *Genética en Medicina*. 2016 (8ª Ed). Elsevier España. ISBN: 9788445826423.
- OMIM (Online Mendelian Inheritance In Man). Es un catálogo continuamente actualizado de los genes humanos y los trastornos genéticos.
<http://www.ncbi.nlm.nih.gov/entrez/Omim>
- 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, ISBN 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).

Additional

- CARRACEDO, A. *Forensic DNA Typing Protocols*. 2005. Humana Press. eISBN: 1-59259-867-6.
- 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.
- EuroGentest. Ofrece información sobre diagnóstico y consejo genético.
<http://www.eurogentest.org/web/index.xhtml>.
- GeneCards. The Human Gene database. Es una base de datos integradora y de búsqueda que proporciona información completa y fácil de usar sobre todos los genes humanos anotados y predichos. Integra automáticamente datos centrados en los genes de ~ 150 fuentes web, incluida información genómica, transcriptómica, proteómica, genética, clínica y funcional.
<http://www.genecards.org>
- LORENTE, J.A. *Un detective llamado ADN*. 2004, Ediciones Temas de Hoy. ISBN: 84-8460-386-5.
- Orphanet <http://www.orpha.net/consor/cgi-bin/index.php>.
Portal de información europeo de referencia en enfermedades raras y medicamentos huérfanos.
- Relación de libros electrónicos sobre temas de biomedicina: <http://www.ncbi.nlm.nih.gov/books>.
- The metabolic and inherited basis of inherited disease, <http://www.ommbid.com>
(acceso libre solo para el resumen de cada patología)



ADDENDUM COVID-19

This addendum will only be activated if the health situation requires so and with the prior agreement of the Governing Council

English version is not available

