# COURSE DATA

| Data Subject | |
|---|---|
| **Code** | 33065 |
| **Name** | Human genetics |
| **Cycle** | Grade |
| **ECTS Credits** | 5.0 |
| **Academic year** | 2017 - 2018 |

**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. Human Genome Project and Biomedicine. Genetic and hereditary diseases

### 2. Human Karyotype

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. Sex chromosomes

Dosage compensation. X-chromosome inactivation: molecular aspects. Abnormalities in the heterochromosomes. Fragile sites. Some errors in gonadal and sexual development

### 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. Genetic 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 and adoption studies. Congenital malformations. Multifactorial disorders of adult life.

### 6. Cloning disease genes

Isolating Mendelian genes: functional, positional and candidate genes cloning. Linkage analysis. Identifying genetic risk factors: association studies.

### 7. Genetic identity

Using molecular markers to obtain genetic fingerprinting. DNA profiling as a tool to assist in the identification of individuals. Assigning probabilities in forensic genetics. Assigning probabilities in paternity testing.

## 8. Biochemical Genetics

Inborn errors of metabolism. Hemoglobinopathies. Thalassemia. Blood groups and incompatibilities

## 9. Genetics of cancer

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

## 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. Pharmacogenetics and personalized medicine. Ethical issues in human genetics.

## 12. Practical sessions

PRACTICE 1. Detecting polymorphisms in candidate genes for alcoholism using synthetic oligonucleotides.
Genomic DNA isolation from buccal mucosa cells. DNA amplification by PCR. Making a dot blot. ASO probe hybridization. Detection of hybrids by colorimetry
PRACTICE 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.
PRACTICE 3. Problems and case studies
Inheritance patterns and pedigree analysis. Genetic risk and recurrence risk assessments. Linkage analysis. 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.** One-hour sessions will be needed to cover this teaching activity, developed in the format of lectures. 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 and problems practical classes.** 5 four-hours sessions and 1 two-hour session will de held. Attendance is compulsory for laboratory classes.

**3. Group mentoring**. In two sessions, one hour each distributed along the course, those concepts that are more complex or more difficult for students will be reviewed.

**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 lasting 1 hour and 15 minutes. The value of this test is 55% 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 forty-five minutes 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 of half-hour (15% of final grade) to be held in conjunction with the previous examinations. The value of the practical skills is 45% 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 will be saved for the second call if the mark is equal to or greater than 5 out of 10. The grade obtained from the practical skills is saved for the second call and 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.

## REFERENCES

### Basic

- GARDNER and SUTHERLAND. Chromosome Abnormalities and Genetic Counseling. 2003 (3ªed) Oxford Monographs on Medical Genetics, Oxford University Press ISBN: 0195149602
- JORDE, CAREY, BAMSHAD y WHITE. Medical genetics. 2003 (3ªed). Hay traducción al castellano: Genética Médica, 2004. Editorial Elsevier España. ISBN 978-84-8174-763-8.
- LEWIS R. Human genetics: Concepts and applications. 2002 (5ª ed). McGraw-Hill. ISBN: 007246268X
- NOVO VILLAVERDE F.J. Genética humana: conceptos, mecanismos y aplicaciones de la genética en el campo de la biomedicina: texto multimedia. 2007. Diaz de Santos. ISBN: 978-84-832-2359-8
- NUSSBAUM R.L., McINNES R.R., WILLARD H.F. Genética en Medicina. 2008 (7ª Ed). Elsevier España. ISBN: 978-84-458-1225-9
- STRACHAN and READ. Human Molecular Genetics. 1996 (1ª ed); 1999 (2ª ed.). BIOS Scient. Publ. 2004 (3ª ed) Garland Science/Taylor & Francis Group. La primera y tercera edición tiene traducción al castellano (1999; 2006)
- SOLARI A.J. Genética humana: fundamentos y aplicaciones en medicina. 2004. Editoral Diaz de Santos ISBN 978-84-7903-926-4
- 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

- PETER TURNPENNY. Emery, Elementos de Genética Médica. 2009 (13ª ed). Elsevier-Masson: Saunder, Mosby, Harcourt Brace

**Additional**

- BUTLER, J.M. Forensic DNA Typing. 2005 (2ª ed). Elsevier. ISBN-13: 978-0-12-147952-7
- CARRACEDO, A. Forensic DNA Typing Protocols. 2005. Humana Press. eISBN: 1-59259-867-6
- JOBLING, HURLES, TYLER-SMITH. Human Evolutionary Genetics: Origins, Peoples & Disease. 2003 Garland Science/Taylor & Francis Group. ISBN: 0815341857
- LORENTE, J.A. Un detective llamado ADN. 2004 Ediciones Temas de Hoy. ISBN: 84-8460-386-5
- MULLER and YOUNG. Emery's Elements of Medical Genetics. 2002 (11ªed). W B Saunders. ISBN: 044307125X
- SUDBERY. Human molecular genetics. 2002 (2ªed). Pearson Education. ISBN: 0130428116
- FRED B. Principles of Cancer Genetics. 2008. Springer Science+Business Media B.V. ISBN 978-1-4020-6783-9.
- Orphanet http://www.orpha.net/consor/cgi-bin/index.php.
  Portal de información europeo de referencia en enfermedades raras y medicamentos huérfanos
- SCRIVER, A.L., BEAUDET, W.S. SLY and D.VALLE (eds). The metabolic and molecular bases of inherited disease, 2001(8ª ed) McGraw-Hill.