Universitat de Lleida

DEGREE CURRICULUM

HUMAN GENETICS

Coordination: FIBLA PALAZON, JUAN

Academic year 2023-24

HUMAN GENETICS 2023-24

Subject's general information

<table>
<tr><td>Subject name</td><td>HUMAN GENETICS</td></tr>
<tr><td>Code</td><td>101529</td></tr>
<tr><td>Semester</td><td>1st Q(SEMESTER) CONTINUED EVALUATION</td></tr>
<tr><td>Typology</td><td>
<table>
<tr><th>Degree</th><th>Course</th><th>Character</th><th>Modality</th></tr>
<tr><td>Bachelor's Degree in Biomedical Sciences</td><td>4</td><td>OPTIONAL</td><td>Attendance-based</td></tr>
<tr><td>Master's Degree in Biomedical Research</td><td></td><td>COMPLEMENTARY TRAINING</td><td>Attendance-based</td></tr>
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<tr><td>Course number of credits (ECTS)</td><td>6</td></tr>
<tr><td>Type of activity, credits, and groups</td><td>
<table>
<tr><th>Activity type</th><th>PRALAB</th><th>PRAULA</th><th>TEORIA</th></tr>
<tr><td>Number of credits</td><td>1</td><td>1</td><td>4</td></tr>
<tr><td>Number of groups</td><td>2</td><td>1</td><td>1</td></tr>
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<tr><td>Coordination</td><td>FIBLA PALAZON, JUAN</td></tr>
<tr><td>Department</td><td>BASIC MEDICAL SCIENCES</td></tr>
<tr><td>Important information on data processing</td><td>Consult this link for more information.</td></tr>
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HUMAN GENETICS 2023-24

Teaching staff	E-mail addresses	Credits taught by teacher	Office and hour of attention
FIBLA PALAZON, JUAN	joan.fibla@udl.cat	0	
FIBLA PALAZON, JUAN	joan.fibla@udl.cat	3	
LOPEZ ORTEGA, RICARDO ENRIQUE	ricard.lopez@udl.cat	2,7	
MONTAL ROURA, ROBERT	robert.montal@udl.cat	1,3	

Learning objectives

The student who passes the course must: (Knowledge objectives)

- Know the terminology and basic vocabulary of human genetics.
- Know the singularities of genetic analysis and its application in Human Genetics.
- Know the human chromosomal structure and the bases of the main chromosomal diseases.
- Know the genetic bases of the most significant monogenic diseases.
- Understand the origin of genetic variability and its application in determining genetic predisposition to diseases.
- Know the basic statistical methods and tools for application in Human Genetics studies
- Know the main human genome databases. Know the genetic diagnosis technologies.

The student who passes the course must be able to: (Ability objectives)

- Correctly interpret the genealogical trees of human families.
- Propose hypotheses about inheritance patterns in human families based on the results or available experimental data.
- Obtain information and correctly interpret information from databases and bibliographic resources in Human Genetics.
- Correctly calculate the genetic risk applying the statistical tools and diagnostic methodologies available.
- It is capable of conducting a genetic association study based on a case / control model.
- Approach problem solving with critical judgment incorporating available scientific information on the subject.
- Develop skills in laboratory work, applying quality criteria and good practice. Develop oral and written communication skills of scientific results.
- Apply genetic analysis in the study and characterization of human diseases.
- Describe the role of individual and population genetic factors in maintaining health and preventing disease
- Apply genetic, chromosomal and molecular diagnostic techniques

Competences

CE1. Apply and formulate mathematical and physical concepts of relevance for the study of human biology.

CE2. Apply the mathematical and physical concepts learned in biomedical experiments and research.

Subject contents

Module 1 Human Genetics

- Unit 1. - Organization of the human genome. Genomic architecture and inheritance.
- Unit 2. - Applications of genetic analysis. Determination of the genetic basis of human phenotypes. Concept of penetration of a character.
- Unit 3 .- Genetic analysis in the study of diseases. Identification of genes involved in human pathologies. Binding and association studies. Case / control design. Global genome analysis (WGA) and candidate genes.

Module 2 Medical Genetics

- Unit 4.- Genomic diseases. Numerical and structural chromosomal alterations. Chromosomopathies. Down syndrome. Genomic syndromes by microdeletions, duplications and inversions. Segmental duplications and CNV.
- Unit 5.- Gene Mutation. Somatic / germinal mutations. Function gain / loss mutations. Mutational models and genetic heterogeneity.
- Unit 6.- Somatic mutation and cancer. Nothing involved in the development of cancer. Oncogenes-Genes tumor suppressors-Genes repair.

Module 3 Clinical Genetics

- Unit 7.- Methodologies of genetic diagnosis. Chromosomal diagnosis. Techniques based on the use of labeled probes (FISH). Genome Comparative Analysis (CGH).
- Unit 8.- Molecular diagnosis, applications. Standardization of molecular diagnostic techniques.
- Unit 9.- Genetic advice. Risk calculation. Module 4.- Human Genomics
- Unit 10.- Mass sequencing. Analysis and interpretation of sequencing data. Applications in genetic diagnosis.

Module 5.- Seminar sessions.

- Preparation / presentation of seminars on human genetic pathologies. Elaboration of a work of revision and bibliographic compilation on certain cases or pathologies. (Each group will have to present a summary of the work in 2-3 sheets DIN A4 and make an oral presentation lasting 25 min)

Module 6.- Practical activities / problems

- Laboratory Sessions
  - LABORATORY PRACTICE - Genetic Diagnostic Laboratory
- Computer Classroom
  - COMPUTING 1 - Consultation and management of databases of interest in Human Genetics.
  - COMPUTING 2, 3 - Statistical resources. Case / control design. Discussion and resolution of a case study.
- Problem sessions. Problem solving and exercises in class.

Methodology

	Activity Summary description of the activity (Subject title or practical activity)	Student face-to-face dedication (hours)	Groups	Total dedication of teachers (hours)
TEO	Theory	**30x1h=30h**	**1**	**30**
LAB	**Laboratory**	**5x2h=10h**	**1**	**10**

INF	Online applications and statistical resources	**3x2h=6h**	**1**	**6**
PROB **SEM**	Case study and problems Seminars (student exhibition)	**6x1h=6h** **4x2h=8h**	**1** **2**	**6** **16**
TOTALS		**60**		**68**

Evaluation

Assessment of learning		
	% nota final	**Type of evaluation**
Theory	75	Final written test on contents and concepts presented in class sessions, seminars and problems.
Practices	5	Assistance and participation in practical activities.
Exercises	5	Problem solving in class
Homework	15	Written summary presentation and class presentation of a model genetic pathology

Bibliography

References

- Sudbery P. (2004). Genética molecular humana. Pearson/Prentice Hall.
- Nussbaum RL, et al. (2004). Genetics in Medicine. Thompson&Thompson.
- Strachan, T, Andrew P Read, and T Strachan (2011). *Human molecular genetics*. New York: Garland Science.
- Gluckman, P., Beedle, A., Hanson, M., (2009). Principles of Evolutionary Medicine, 1st ed. Oxford University Press, USA.
- Young, I.D., (2006). Introduction to Risk Calculation in Genetic Counseling, 3rd ed. Oxford University Press, USA.
- Turnpenny, Ellard, (2011). Emery's Elements of Medical Genetics: With STUDENT CONSULT Online Access, 14e, 14th ed. Churchill Livingstone

NCBI on-line books

- **GeneReviews™** Edited by Roberta A Pagon, Editor-in-chief, Thomas D Bird, Cynthia R Dolan, Karen Stephens, and Margaret P Adam. **http://www.ncbi.nlm.nih.gov/books/NBK1116/**
- **Human Molecular Genetics, 2nd edition** Tom Strachan and Andrew P Read. http://www.ncbi.nlm.nih.gov/books/NBK7580/