



Teaching Guide

Identifying Data					2020/21
Subject (*)	Human Genetics	Code	610441016		
Study programme	Mestrado Universitario en Bioloxía Molecular , Celular e Xenética				
Descriptors					
Cycle	Period	Year	Type	Credits	
Official Master's Degree	2nd four-month period	First	Optional	3	
Language	Spanish				
Teaching method	Face-to-face				
Prerequisites					
Department	Bioloxía				
Coordinador	Gonzalez Tizon, Ana Maria	E-mail	ana.gonzalez.tizon@udc.es		
Lecturers	Gonzalez Tizon, Ana Maria Martinez Lage, Andres	E-mail	ana.gonzalez.tizon@udc.es andres.martinez@udc.es		
Web					
General description	Esta materia estudia la organización, estructura y función del genoma humano, profundizando en el conocimiento de las enfermedades genéticas humanas e identificación de individuos. Se abordan y tratan las técnicas actuales de análisis genómico para el estudio, aislamiento y cartografía de genes y de diagnóstico molecular.				
Contingency plan	<ol style="list-style-type: none"> 1. Contents will be the same. 2. In-person instruction will change to virtual-only. This means that all lectures will be hosted using MS TEAMS. 3. Tutoring sessions and any other communication will take place by means of email, videocalls or chat as implemented in MS TEAMS. 4. The only change in the assessment will be that all students will be evaluated online. 5. The recommended reference list will remain the same. If needed, instructors will provide with any reading and/or course resources to the students. 				

Study programme competences

Code	Study programme competences
A1	Skills of working in a sure way in the laboratories knowing operation handbooks and actions to avoid incidents of risk.
A6	Skills of understanding the functioning of cells through the structural organization, biochemistry, gene expression and genetic variability.
A8	Skills of having an integrated view of the previously acquired knowledge about Molecular and Cellular Biology and Genetics, with an interdisciplinary approach and experimental work.
A11	Skills of understanding the structure, dynamics and evolution of genomes and to apply tools necessary to his study.
A12	Skills to understand, detect and analyze the genetic variation, knowing genotoxicity processes and methodologies for its evaluation, as well as carrying out diagnosis and genetic risk studies.
B1	Analysis skills to understand biological problems in connection with the Molecular and Cellular Biology and Genetics.
B3	Skills of management of the information: that are able to gather and to understand relevant information and results, obtaining conclusions and to prepare reasoned reports on scientific and biotechnological questions
B5	Correct oral and written communication on scientific topics in the native language and at least in another International diffusion language.
B6	Skills of team work: that are able to keep efficient interpersonal relationships in an interdisciplinary and international work context, with respect for the cultural diversity.
B8	Critical reasoning skills and ethical commitment with the society: sensitivity in front of bioethical problems and to the ones related to the natural resource conservation
C1	Adequate oral and written expression in the official languages.

Learning outcomes



Learning outcomes	Study programme competences		
Capacidad de realizar análisis genéticos tanto a nivel molecular como en la identificación de enfermedades genéticas mediante estudios familiares.	AR1	BR1	CC1
Capacidad de realizar diagnóstico genético.	AR6	BR3	
	AR8	BR5	
	AR11	BR6	
	AR12	BR8	

Contents	
Topic	Sub-topic
Lecture 1. THE HUMAN GENOME: SEQUENCE AND VARIATION	Functional elements Protein-coding genes Non-coding, RNA-only genes Repetitive elements Mitochondrial genome Genomic variability Epigenetics
LECTURE 3. CHROMOSOMES AND CLINICAL SYNDROMES	The human karyotype Mitotic and meiotic alterations: non-disjunction Changes in Number and structure of the chromosomes Mosaics
LECTURE 3. GENES AND CANCER	Oncogenes and tumor supressor genes Germline mutations: familiar cancer Somatic cancer genetics
LECTURE 4. FORENSIC GENETICS	DNA fingerprinting
LECTURE 5. EVOLTION OF HUMAN POPULATIONS	Genetic diversity Mitochondial and nuclear inheritance
LECTURE 6. GENETIC DISEASES	Familiar studies Mendelian inheritance Multiple alleles Complex inheritance Anticipation, expressivity, penetrance, mosaicism, mitochondrial inheritance and dynamic mutations hEREDABILITY
LABORATORY PRACTICES	1- Pedigrees 2- Genetic polymorphism analyses 3- Phylogenies

Planning				
Methodologies / tests	Competencies	Ordinary class hours	Student?s personal work hours	Total hours
Guest lecture / keynote speech	A6 A11 A12	12	18	30
Laboratory practice	A1 A8 B1 B3 B5 B6 B8	14	7	21



Mixed objective/subjective test	B1 B5	2	0	2
Practical test:	A6 B1 B3 C1	2	0	2
Online discussion	A6 A8 A11 B3 B5	2	14	16
Personalized attention		4	0	4

(*)The information in the planning table is for guidance only and does not take into account the heterogeneity of the students.

Methodologies	
Methodologies	Description
Guest lecture / keynote speech	In each class, contents related to different aspects of the syllabus will be exposed. The teacher will explain the fundamental contents of each topic and indicate the activities associated with it.
Laboratory practice	The practical classes will consist of an explanation on the part of the professor on the conceptual bases and the objectives to reach and the development of tasks on the part of the student. It is intended that the student has maximum autonomy, facilitating means and guidance.
Mixed objective/subjective test	Written test in which any aspect addressed in both theoretical and practical teaching will be discussed.
Practical test:	
Online discussion	Students will read one or two recent articles on genetic diseases and prepare a presentation that they will defend in a time of 10-12 minutes. The consulted bibliography will be valued, synthesis capacity, oral expression and argumentation. In the case of non-contact students, they must do the same activity, substituting the oral presentation for a written work of no more than 4 pages.

Personalized attention	
Methodologies	Description
Mixed objective/subjective test	Alumnado PRESENCIAL: O alumnado poderá acudir as tutorías dos profesores nos horarios previamente establecidos ou consensuados cos alumnos e alumnas da materia. Alumnado SEMIPRESENCIAL: solicitará tutoría e será atendido vía correo electrónico ou plataforma Moodle.
Guest lecture / keynote speech	PLAN DE CONTINXENCIA: en caso de confinamento o alumnado solicitará tutoría e se fará vía TEAMS ou correo electrónico, tanto para o alumnado PRESENCIAL como SEMIPRESENCIAL.
Laboratory practice	
Online discussion	
Practical test:	

Assessment			
Methodologies	Competencies	Description	Qualification
Mixed objective/subjective test	B1 B5	The domain of theoretical and practical concepts will be valued, clarity in the explanations, capacity to relate and integrate the information received treated in the magisterial sessions and in the laboratory and bioinformatics practices, and capacity to solve questions and problems.	40
Laboratory practice	A1 A8 B1 B3 B5 B6 B8	It will be valued the knowledge about the meaning of the tasks carried out, and the interpretation of the results obtained.	15
Online discussion	A6 A8 A11 B3 B5	Students will prepare a 10-minute presentation on a genetic disease. The students will not be present and will do the power point activity as well as the written memory of the work.	30
Practical test:	A6 B1 B3 C1	The knowledge acquired during the laboratory practices will be valued.	15



Assessment comments

Se considerará NO PRESENTADO cuando el estudiante no haya realizado NINGUNA de las actividades/metodologías propuestas.

Sources of information

Basic	<ul style="list-style-type: none"> - Strachan, T. & Read, A.P. (2004). Genética Molecular Humana (3ª ed). McGrawHill, México. - Pasternak, Jack (2005). An introduction to human molecular genetics. Hoboken, New Jersey. John Wiley & Sons - T Strachan, AP Read (2010). Human Molecular Genetics 4th ed.. Garland Science
Complementary	<ul style="list-style-type: none"> - Emery, A.E.H. & Mueller, R.F. (1992). Principios de Genética Médica.. Churchill Livingstone. - Jorde, L.B. Carey, J.C. & White, R.L. (1996). Genética Médica.. Mosby. - Novo Villaverde, F.J. (2007). Genética humana. Conceptos, mecanismos y aplicaciones de la Genética en el campo de la biomedicina. . Pearson, Prentice Hall. Madrid. - Sudbery, P. (2004). Genética molecular humana. . Pearson, Prentice Hall. 2ª ed. Madrid. - Jobling, M.A.; Hurler, M.E. ; Tyler-Smith, C. (2004). Human evolutionary genetics: origins, peoples & disease. New York, Garland Publishing - Maroni, G. (2001). Molecular and Genetic Analysis of Human Trait.. Blackwell Science. Malden, MA, USA. - Vogel, F. & Motulsky, A.G. (1997). Human Genetics: Problems and Approaches (3th ed). Springer Verlag, Heidelberg, Germany - Cummings, Michael R. (2003). Human heredity: principles and issues. Pacific Grove, California. Thompson - King, Roger (2000). Cancer biology (2º ed). Essex, UK. Pearson Education Limited - McKinnell R.; Parchment, R. et al (2006). The biological basis fo cancer (2º ed). Cambridge, NY. Cambridge University Press - Pecornio, Lauren (2005). Molecular biology of cancer. Oxford, UK. Oxford University Press <p>O alumnado PRESENCIAL E SEMIPRESENCIAL, e recibirá por parte dos profesores da materia webgrafía recente e artigos de revisión para preparar axeitadamente a materia.PLAN DE CONTIXENCIA: se aplicará o mesmo tratamento (proporcionaremos webgrafía axeitada) na plataforma Moodle.</p>

Recommendations

Subjects that it is recommended to have taken before

Subjects that are recommended to be taken simultaneously

Immunology/610441008
Stem Cells and Cell Therapy/610441009

Subjects that continue the syllabus

Cellular Techniques/610441001
Molecular Techniques/610441002
Genetic Variation Mechanisms/610441005

Other comments

(*The teaching guide is the document in which the URV publishes the information about all its courses. It is a public document and cannot be modified. Only in exceptional cases can it be revised by the competent agent or duly revised so that it is in line with current legislation.