Teaching Guide				
Identifying Data				2019/20
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.			

Study programme competences / results	
Code	Study programme competences / results
A2	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 / results		
Capacidad de realizar análisis genéticos tanto a nivel molecular como en la identificación de enfermedades genéticas mediante estudios familiares. Capacidad de realizar diagnóstico genético.	AR2 AR6 AR8 AR11 AR12	BR1 BR3 BR5 BR6 BR8	CC1

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-disyunction 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 / Results	Teaching hours (in-person & virtual)	Student?s personal work hours	Total hours
Guest lecture / keynote speech	A6 A11 A12	12	18	30
Laboratory practice	A2 A8 B1 B3 B5 B6 B8	14	7	21
Mixed objective/subjective test	B1 B5	2	0	2
Online discussion	A6 A8 A11 B3 B5	2	16	18
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.
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 Guest lecture / keynote speech Laboratory practice Online discussion	No existe ningún límite en el número de hoara determinado a tutorias. Los estudiantes podrán acudir a tutorias de los profesores en aquellos horarios previamente establecidos en el primer apartado.

Assessment			
Methodologies	**Competencies / Results**	**Description**	**Qualification**
Mixed objective/subjective test	B1 B5	Se valorará el dominio de conceptos teóricos y prácticos, claridad en las explicaciones, capacidad de relacionar e integrar la información recibida tratada en las sesiones magistrales y en las prácticas de laboratorio y bioinformática, y cpacidad de resolver cuestiones y problemas. Se evaluarán las competencias específicas A3, A9 y A11	40
Laboratory practice	A2 A8 B1 B3 B5 B6 B8	Se valorará el conocimiento sobre el significado de las tareas realizadas, y la interpretación de los resultados obtenidos. Se evaluarán las competencias específicas A3 y A4	30
Online discussion	A6 A8 A11 B3 B5	Os alumnos preparán unha presentación de 10 minutos sobre una enfermidade xenética. Os alumnos non presenciais farán dita actividade facendo tanto o power point como a memoria escrita do traballo.	30

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

Sources of information

Basic	- 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 .O {font-size:149%;}.O {font-size:149%;}
Complementary	- 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. (2004). Genética molecular humana. . Pearson, Prentice Hall. 2ª ed. Madrid. - Jobling, M.A.; Hurles, M.E. ; Tyler-Smith, C. (2004). Human evolutionary genetics: origins, peolples & disease. New York, Garland Plublishing - 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 .O {font-size:149%;}.O {font-size:149%;}

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.