Teaching Guide				
Identifying Data				2017/18
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	Optativa	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	
Code	Study programme competences
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

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. Capacidad de realizar diagnóstico genético.	AR2 AR6 AR8 AR11 AR12	BR1 BR3 BR5 BR6 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-disyunction Changes in Number and structure of the chromosomes Mosaics
LECTURE 4. MODES OF INHERITANCE	Familiar studies Mendelian inheritance Multiple alleles Complex inheritance Anticipation, expressivity, penetrance, mosaicism, mitochondrial inheritance and dynamic mutations
TEMA 4. ENFERMEDADES POLIXÉNICAS E MULTIFACTORIAIS.	Polixenes e variacións no fenotipo. Heredabilidade.
LECTURE 5. GENES AND CANCER.	Oncogenes and tumor supressor genes Germline mutations: familiar cancer Somatic cancer genetics
LECTURE 6. GENE THERAPY	Somatic and germinal gene therapy. Gene therapy vectors and nonviral vectors Ex vivo and in vivo methods.
LABORATORY PRACTICES	1. The human karyotype and the identification of chromosome alterations. 2. Bioinformatics: search and comparison of DNA sequences. Databases and genome browsers

Planning				
Methodologies / tests	Competencies	Ordinary class hours	Student?s personal work hours	Total hours
Guest lecture / keynote speech	A6 A11 A12	14	21	35
Laboratory practice	A2 A8 B1 B3 B5 B6 B8	14	7	21
Student portfolio	A6 A8 A11 B3 B5	0	13	13
Mixed objective/subjective test	B1 B5	2	0	2
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	En cada clase se expondrán contenidos relacionados con diferentes aspectos del temario. El profesor explicará los contenidos fundamentales de cada tema y señalará las actividades asociadas al mismo.
Laboratory practice	Las clases prácticas consistirán de una explicación por parte del profesor sobre las bases conceptuales y los objetivos a alcanzar y el desarrollo de tareas por parte del alumno. Se pretende que el alumno tenga la máxima autonomía, facilitándole medios y orientación.
Student portfolio	Los estudiantes contestarán a unas fichas que les serán entregadas por los profesores sobre diferentes aspectos teóricos y prácticos de la materia.
Mixed objective/subjective test	Prueba escrita en la que se tratará cualquier aspecto abordado en la docencia tanto teórica como práctica.

Personalized attention	
Methodologies	**Description**
Student portfolio Mixed objective/subjective test Guest lecture / keynote speech Laboratory practice	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**	**Description**	**Qualification**
Student portfolio	A6 A8 A11 B3 B5	Se valorará el grado de comprensión, análisis, calidad y claridad en las respuestas y el tratamiento de las cuestiones y problemas planteados. Se evaluarán las competencias específicas A3, A9 y A11	30
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

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.