Teaching Guide				
Identifying Data				2023/24
Subject (*)	Human Genetics		Code	610441017s
Study programme	Máster Universitario en Bioloxía Molecular, Celular e Xenética (semipresencial)			
Descriptors				
Cycle	Period	Year	Type	Credits
Official Master's Degree	2nd four-month period	First	Optional	3
Language	Spanish			
Teaching method	Hybrid			
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	This subject studies the organization, structure and function of the human genome, deepening the knowledge of human genetic diseases and identification of individuals. Current genomic analysis techniques for the study, isolation and mapping of genes and molecular diagnosis are addressed and discussed.			

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	Ability to draft, represent, analyze, interpret and present technical documentation and relevant data in the field of the branch of knowledge of the master's degree 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
C2	Ability to know and use appropriately the technical terminology of the field of knowledge of the master, in the native language and in English, as a language of international diffusion in this field
C9	Ability to manage times and resources: developing plans, prioritizing activities, identifying critical points, establishing goals and accomplishing them.

Learning outcomes			
Learning outcomes	Study programme competences		

Capacidade de realizar análise xenéticos tanto a nivel molecular como na identificación de enfermedades xenéticas mediante estudos familiares. Capacidade de realizar diagnóstico xenético.	AR1 AR6 AR8 AR11 AR12	BR1 BR3 BR5 BR6 BR8	CC2 CC9

Contents	
Topic	**Sub-topic**
TEMA 1. O XENOMA HUMANO: secuencia e variación.	Elementos funcionais Xenes que codifican para proteínas Xenes que codifican para RNAs Elementos repetitivos Xenoma mitocondrial Variabilidade xenética Epixenética
TEMA 2. CROMOSOMAS E ALTERACIONS CROMOSÓMICAS	Cariotipo humano Alteracions mitóticas e meióticas: non disyunción. Alteracions cromosómicas numéricas e estructurais. Mosaicismo Enfermedades producidas por alteracions cromosómicas
TEMA 3. XENES e CÁNCER.	Oncoxenes e xenes supresores de tumores. Xenética de cánceres comunes. Cánceres familiares.
Tema 4. XENÉTICA FORENSE	Identificación xenética de individuos. Pegada xenética.
TEMA 5. EVOLUCIÓN DAS POBOACIÓNS HUMANAS	Diversidade xenética humana Variacions poboacionais Herdanza mitocondrial Herdanza nuclear
TEMA 6. ENFERMIDADES	Herdanza mendeliana. Factores que complican os patrons de herencia. Polixenes e variacións no fenotipo. Heredabilidade.
TEMARIO DE PRÁCTICAS DE LABORATORIO	1-Análise de haplotipos mitocondriais 2-Filoxenias humanas.

Planning				
Methodologies / tests	**Competencies**	**Ordinary class hours**	**Student?s personal work hours**	**Total hours**
Mixed objective/subjective test	A1 A6 A11 B1 B3 B6 B8	0	2	2
Practical test:	A1 A11 B1 B5 B6 B8	0	3	3
ICT practicals	A8 A12 B5 B8 C2 C9	4	6	10
Workbook	A8 A11 A12 B1 B3 B5	1	14	15
Guest lecture / keynote speech	A6 A8 A11 A12	0	41	41
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
Mixed objective/subjective test	Proba escrita na que se evaluará cualquier aspecto relacionado coas clases expositivas da materia.
Practical test:	Os estudantes realizarán exercicios similares os que fixeron durante as prácticas TIC
ICT practicals	As prácticas presenciais obligatorias de laboratorio serán sustituídas por prácticas virtuales que serán presenciais via telemática.
Workbook	Os estudantes deberán leer, alo menos dous artígos científicos propostos polo profesor e presentar un resumen de cada uno de eles en formato pdf.
Guest lecture / keynote speech	Engadirache as presentacios da materia, artigos científicos e links a páxinas web. Durante as 4 horas de tutorias presenciais os estudantes consultarán as dúbidas que teñan.

Personalized attention	
Methodologies	**Description**
ICT practicals Guest lecture / keynote speech	Os alumnos deberán manter catro horas de tutorías presenciais co profesor para resolución de dúbidas vía TEAMS. Ademais poderase solicitar outras tutorias ou ben consulta de dúbidas via correo electrónico ou chat implementado en TEAMS en horario a convir por ambas partes.

Assessment			
Methodologies	**Competencies**	**Description**	**Qualification**
ICT practicals	A8 A12 B5 B8 C2 C9	Se valorará el conocimiento sobre el significado de las tareas realizadas, y la interpretación de los resultados obtenidos.	15
Workbook	A8 A11 A12 B1 B3 B5	Los alumnos realizarán un resumen de 2-3 páginas que deberán entregar en formato pdf a los profesores, tras la lectura de dos artículos de investigación. Se valorará la expresión escrita, la claridad y concisión, y la capacidad de discusión.	30
Practical test:	A1 A11 B1 B5 B6 B8	rueba escrita en la que se valorarán los conocimientos adquiridos durante las prácticas a través de TICs	15
Mixed objective/subjective test	A1 A6 A11 B1 B3 B6 B8	Se valorará el dominio de conceptos teóricos, claridad en las explicaciones, capacidad de relacionar e integrar la información recibida tratada en las sesiones magistrales y en las prácticas a través de TICs, y capacidad de resolver cuestiones y problemas.	40

Assessment comments

Las prácticas a través de TICs son obligatorias. Para aprobar la materia el alumno debe obtene al menos un 50% de la puntuación asignada a la prueba mixta y otro 50% de la de las prácticas. Se considerará NO PRESENTADO cuando el alumno no haya participado en más de un 20% de las actividades evaluables programadas. Este criterio se aplica a la convocatoria de junio. En la convocatoria de julio, para obtener la calificación NO PRESENTADO, bastará con no presentarse a las pruebas objetivas (exámenes de teoría y prácticas). Para la evaluación de la convocatoria de julio, el alumno, además de los exámenes de teoría y prácticas, deberá presentar los resúmenes de las lecturas. En el caso de que esta actividad estuviese ya evaluada en la convocatoria anterior, la calificación obtenida se mantendrá para julio.

Sources of information	
Basic	El alumnado tendrá acceso, a través de la plataforma Moodle, a una webgrafía necesaria y suficiente para superar con éxito la asignatura.Esta webgrafía estará disponible a principios de curso de la materia.
Complementary	

Recommendations
Subjects that it is recommended to have taken before
Immunology/610441009 Genetic Variation Mechanisms/610441005
Subjects that are recommended to be taken simultaneously
Cellular Techniques/610441001
Subjects that continue the syllabus
Other comments

Attendance at lectures makes it possible to deal with any doubts or questions that may arise in the course of the explanations, facilitating the understanding of the subjects. Study should include regular reading of at least the recommended bibliography. Group study and work favours understanding and develops a critical spirit. The doubts and difficulties that arise in any aspect of the subject will be resolved as soon as possible, raising them in the classroom or attending individual tutorials. Given that part of the recommended bibliography for this subject is in English, it is recommended to have a good command of this language, at least at the level of comprehension of written texts.Gender PerspectiveIn this subject, the gender perspective will be taken into account, sexist attitudes will not be tolerated and the values of respect and equality will be promoted.Program Green Campus
Empower of SciencesTo help to achieve some sustainable immediate surroundings and fulfil with the point 6 of the Environmental Statement of the faculty of Sciences (2020), the documentary works that realise in this matter:a. They will request mostly in virtual format and computer supportb. To realise in paper:-they will not employ plastic-will realise impressions to double expensive-will employ paper recycled-will avoid the realisation of draftsTo Environmental Statement is available in:https://ciencias.udc.es/images/Facultade/Green_Campus/Regulamento_Comit%C3%A9_Green_Campus_FCiencias.pdf