Teaching Guide				
Identifying Data				2022/23
Subject (*)	Human Genetics		Code	610441017
Study programme	Máster 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	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 / results	
Code	Study programme competences / results
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
C1	Ability to express oneself correctly, both orally and in writing, in the official languages of the autonomous community
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 / results		
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	CC1 CC9

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	A8 A11 A12 B1 B8	12	18	30
Laboratory practice	A1 A6 B3 B5 B6 C1 C9	14	7	21
Mixed objective/subjective test	A1 A5 A7 A11 B1 B2 B3 B4 B6 B7 B8 B9 B10 B12 B13 C1 C3 C4 C6 C7 C8	2	0	2
Practical test:	A1 A5 A7 A11 B1 B2 B3 B4 B5 B6 B8 B9 B10 B12 B13 C1 C3 C4 C6 C7 C8	2	0	2

Oral presentation	A1 B1 B2 B3 B4 B5 B6 B7 B8 B9 B10 B11 B12 B13 C1 C3 C4 C6 C7 C8	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:	A written test will be carried out to evaluate the knowledge acquired during the laboratory practices
Oral presentation	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.

Personalized attention	
Methodologies	**Description**
Mixed objective/subjective test Guest lecture / keynote speech Laboratory practice Oral presentation Practical test:	Students: will request tutoring and will be attended via email or TEAMS. CONTINXENCE PLAN: in case of confinement, students will request tuition and it will be done via TEAMS or email

Assessment			
Methodologies	**Competencies / Results**	**Description**	**Qualification**
Mixed objective/subjective test	A1 A5 A7 A11 B1 B2 B3 B4 B6 B7 B8 B9 B10 B12 B13 C1 C3 C4 C6 C7 C8	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 A6 B3 B5 B6 C1 C9	It will be valued the knowledge about the meaning of the tasks carried out, and the interpretation of the results obtained.	15
Oral presentation	A1 B1 B2 B3 B4 B5 B6 B7 B8 B9 B10 B11 B12 B13 C1 C3 C4 C6 C7 C8	Students will prepare a 10-minute presentation on a topic related to any of the content covered in the subject.	30

Practical test:	A1 A5 A7 A11 B1 B2 B3 B4 B5 B6 B8 B9 B10 B12 B13 C1 C3 C4 C6 C7 C8	The knowledge acquired during the laboratory practices will be valued.	15

Assessment comments
Laboratory practices are mandatory.To pass the subject, the student must obtain at least 50% of the score assigned to the mixed test and another 50% of that of the laboratory practices.It will be considered NOT PRESENTED when the student has not participated in more than 20% of the scheduled assessable activities. This criterion applies to the June call. In the July call, to obtain the grade NOT PRESENTED, it will be enough to not appear for the objective tests (theory and practical exams).For the evaluation of the July call, the student, in addition to the theory and practical exams, must present the power point presentation of the oral presentation. In the event that this activity was already evaluated in the previous call, the grade obtained will remain for July.For students with part-time dedication and exemption from attendance, the teacher will adopt the measures that he deems appropriate to avoid damaging her grade (flexibility in the delivery dates of the assessable activities). Instead of the oral presentation, these students will make a 2-3 page summary that must be delivered in pdf to the teacher for evaluation. The fraudulent performance of the tests or evaluation activities will directly imply the application of the current regulations of the UDC

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 Nesta materia, os profesores recomendarán artigos científicos de revisión, publicados recentemente, para que o alumnado dispoña de bibliografía e referencias actuais sobre cada un dos temas da materia.Os artículos estarán aloxados na plataforma moodle dende o primeiro día de clase.
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 alumnado PRESENCIAL E SEMIPRESENCIAL, e recibirá por parte dos profesores da materia webgrafía reciente e artículos de revisión para preparar axeitadamente a materia.PLAN DE CONTIXENCIA: se aplicará o mesmo tratamento (proporcionaremos webgrafía axeitada) na plataforma Moodle. .O {font-size:149%;}

Recommendations
Subjects that it is recommended to have taken before
Subjects that are recommended to be taken simultaneously
Immunology/610441009 Stem Cells and Cell Therapy/610441010

Subjects that continue the syllabus
Cellular Techniques/610441001
Molecular Techniques/610441002
Genetic Variation Mechanisms/610441005
Other comments
Attendance at the master classes enables the treatment of doubts or questions that may arise in the course of the explanations, facilitating the understanding of the topics. The study must contemplate the habitual consultation of at least the recommended bibliography. Study and group work favors understanding and develops a critical spirit. Doubts and difficulties raised by any aspect of the subject must be resolved as soon as possible, raising them in face-to-face classes or attending individualized tutorials. Given that part of the recommended bibliography for this subject is in English, it is advisable to have knowledge of this language, at least at the level of comprehension of written texts.Green Campus ProgramGreen Campus Program of the Faculty of SciencesTo help achieve a sustainable environment and comply with point 6 of the "Environmental Declaration of the Faculty of Sciences (2020)", the documentary works carried out in this matter:to. They will be requested mainly in virtual format and computer support.b. If done on paper:- Plastics will not be used- They will be printed on both sides.- Recycled paper will be used- Drafts will be avoided.