| Teaching Guide | | | | |
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| 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<br>Martinez Lage, Andres | E-mail | ana.gonzalez.tizon@udc.es<br>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 | 1. Contents will be the same.<br><br>2. In-person instruction will change to virtual-only. This means that all lectures will be hosted using MS TEAMS.<br><br>3. Tutoring sessions and any other communication will take place by means of email, videocalls or chat as implemented in MS TEAMS.<br><br>4. The only change in the assessment will be that all students will be evaluated online.<br><br>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 / results | |
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| 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 | 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 |
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| 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.<br>Capacidad de realizar diagnóstico genético. | AR1<br>AR6<br>AR8<br>AR11<br>AR12 | BR1<br>BR3<br>BR5<br>BR6<br>BR8 | CC1 |

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

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|---|---|---|---|---|
| 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 | |
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| **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.<br><br>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<br>Guest lecture / keynote speech<br>Laboratory practice<br>Online discussion<br>Practical test: | Alumnado PRESENCIAL: O alumnado podrá acudir as tutorías dos profesores noss horarios previamente establecidos ou consensuados cosalumnos e alumnas da materia.<br>Alumnado SEMIPRESENCIAL: solicitará tutoría e será atendido vía correo electrónico ou plataforma Moodle.<br>PLAN DE CONTINXENCIA: en caso de confinamento o alumnado solicitará turoría e se fará vía TEAMS ou correo electrónico, tanto para o alumnado PRESENCIAL como SEMIPRESENCIAL. |

| Assessment | | | |
|---|---|---|---|
| **Methodologies** | **Competencies / Results** | **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 |

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|---|---|---|---|
| Online discussion | A6 A8 A11 B3 B5 | Students will prepare a 10-minute presentation on a genetic disease.<br><br>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 |
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| Se considerará NO PRESENTADO cuando el estudiante no haya realizado NINGUNA de las actividades/metodologías propuestas. |

| Sources of information | |
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| Basic | - Strachan, T. & Read, A.P. (2004). Genética Molecular Humana (3ª ed). McGrawHill, México.<br>- Pasternak, Jack (2005). An introduction to human molecular genetics. Hoboken, New Jersey. John Wiley & Sons<br>- T Strachan, AP Read (2010). Human Molecular Genetics 4th ed.. Garland Science<br>.O {font-size:149%;}.O {font-size:149%;} |
| Complementary | - Emery, A.E.H. & Mueller, R.F. (1992). Principios de Genética Médica.. Churchill Livingstone.<br>- Jorde, L.B. Carey, J.C. & White, R.L. (1996). Genética Médica.. Mosby.<br>- Novo Villaverde, F.J. (2007). Genética humana. Conceptos, mecanismos y aplicaciones de la Genética en el campo de la biomedicina. . Pearson, Prentice Hall. Madrid.<br>- Sudbery, P. 2004. (2004). Genética molecular humana. . Pearson, Prentice Hall. 2ª ed. Madrid.<br>- Jobling, M.A.; Hurles, M.E. ; Tyler-Smith, C. (2004). Human evolutionary genetics: origins, peolples & disease. New York, Garland Plublishing<br>- Maroni, G. (2001). Molecular and Genetic Analysis of Human Trait.. Blackwell Science. Malden, MA, USA.<br>- Vogel, F. & Motulsky, A.G. (1997). Human Genetics: Problems and Approaches (3th ed). Springer Verlag, Heidelberg, Germany<br>- Cummings, Michael R. (2003). Human heredity: principles and issues. Pacific Grove, California. Thompson<br>- King, Roger (2000). Cancer biology (2º ed). Essex, UK. Pearson Education Limited<br>- McKinnell R.; Parchment, R. et al (2006). The biological basis fo cancer (2º ed). Cambridge, NY. Cambridge University Press<br>- Pecornio, Lauren (2005). Molecular biology of cancer. Oxford, UK. Oxford University Press<br>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 |
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| Subjects that it is recommended to have taken before |
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| Subjects that are recommended to be taken simultaneously |
| Immunology/610441008<br>Stem Cells and Cell Therapy/610441009 |
| Subjects that continue the syllabus |
| Cellular Techniques/610441001<br>Molecular Techniques/610441002<br>Genetic Variation Mechanisms/610441005 |
| Other comments |
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