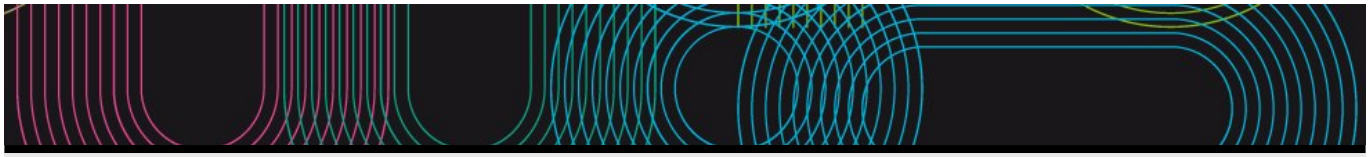

IDENTIFYING DATA				
Human genetics and molecular pathology				
Subject	Human genetics and molecular pathology			
Code	V02G031V01408			
Study programme	Grado en Biología			
Descriptors	ECTS Credits	Choose	Year	Quadmester
	6	Optional	4th	1st
Teaching language	Spanish			
Department				
Coordinator	Valverde Pérez, Diana			
Lecturers	Fernández Silva, Íria Valverde Pérez, Diana			
E-mail	dianaval@uvigo.es			
Web				
General description	This matter will help us to recognise the organisation of the human genome, know and understand the biochemical and genetic changes that occur in different pathologies, study the methodologies used in the diagnostic, follow-up and investigation of illnesses. Matter of the program *English *Friendly: The/ace international students will be able to request to the *profesorado: to) material and bibliographic references for the follow-up of the matter in English, *b) attend the *tutorías in English, *c) proofs and evaluations in English.			

Training and Learning Results	
Code	
A1	Students should prove understanding and knowledge in this study field that starts in the Secundary Education and with a level that, even though it is suppported in advanced books, also includes some aspects that involve knowledge from the vanguard of the study field.
A2	Students should know how to apply their knowledge to their work or vocation in a professional way. They also should have the competences that are usually proved through the elaboration and defence of arguments and the resolution of problems within their study field.
B1	Developing autonomous learning by identifying their own training need and organizing and planning tasks and time.
B2	Manage scientific-technical information using diverse and reliable sources. Analyze data and documents and interpret them critically and rigorously, including considerations on their social relevance and in the professional field of Biology.
B6	Develop analysis and synthesis, critical reasoning and argumentation skills, applying them in Biology and other scientific-technical disciplines.
C3	Perform and interpret molecular, physicochemical and biological analyses, including samples of human origin. Conduct assays and functional tests under normal and abnormal conditions.
C5	Manipulate and analyse genetic material and determine its alterations and pathological implications. Knowing the applications of genetic engineering.
C11	Perform and interpret bioassays, identify chemical and biological agents, including pathogens, as well as their toxic products. Develop and apply biological control techniques.
C12	Writing reports and technical dossiers, as well as directing and executing projects on topics related to biology and its applications.
C16	The ability to identify the genetic and molecular bases of disease, advise on genetic counselling and genomic studies. Understand the control of cellular activity and integrated physiological responses, analysing their repercussions on health.
C17	Understanding the social projection of biology applied to health at its different levels (analytical, pathological and public health) and its repercussions on professional practice.
D5	Communicate effectively and appropriately, including the use of computer tools and English.

Expected results from this subject

Expected results from this subject	Training and Learning Results			
Recognise the organisation of the human genome.	A1 A2	B1 B2 B6	C16	
Know and understand the biochemical and genetic changes that occur in a wide rank of pathologies.	A1 A2	B1 B2 B6	C3 C16	D5
Present the methodologies for the diagnostic, follow-up, and investigation of the illnesses.	A1 A2	B1 B2 B6	C5 C12 C16 C17	D5
Purchase basic skills of laboratory for the diagnostic of illnesses.	A1 A2	B1 B2	C3 C5 C11	D5

Contents	
Topic	
□ The human genome.	Technical analysis of the structure and expression of genes and genomes. Structure of the human genome, genic regulation and epigenome Genetic Variation Genetic of populations and human evolution
□ Citogenética Human.	Chromosomes, cellular division, and human or Technical karyotype of analysis citogenético and clinical diagnostic chromosomal Alterations
□ Genetic base of the human illnesses.	Or Connecting phenotypes and genotypes or Mapping and identification of genes for illnesses *monogénicas
□ Inheritance *multifactorial.	Or Identification of factors of risk and molecular base in complex illnesses or Models of illness of inheritance *multifactorial
□ Genetics of the cancer.	Or genetic and environmental Factors of the cancer or *Oncogenes and genes *supresores or *Epigenética
□ Molecular pathology of human illnesses.	Or *Metabolopatías or *Transtornos *monogénicos or polygenic Inheritance
□ Molecular diagnostic.	Or Technical used or Indications for the proofs or Diagnostic *postnatal, *prenatal and *preimplantatorio or genetic Advice and ethical appearances

Planning	Class hours	Hours outside the classroom	Total hours
Lecturing	30	30	60
Practices through ICT	12	48	60
Case studies	3	6	9
Objective questions exam	1	5	6
Objective questions exam	1	5	6
Case studies	1	1	2
Report of practices, practicum and external practices	0	4	4
Presentation	1	0	1
Problem and/or exercise solving	1	1	2
*The information in the planning table is for guidance only and does not take into account the heterogeneity of the students.			

Methodologies	Description
Lecturing	Exhibition by part of the professor of the foundations and basic principles. Like support to the theoretical explanations, will provide to the educational material students adapted through the platform Moovi of the Virtual Campus
Practices through ICT	To the students will request them the delivery of a report of practice and/or resolution of questions and/or exercises. Like support to practise them, will provide the students with the appropriate educational material through the platform Moovi of the Virtual Campus
Case studies	To the start of the course will deliver them to the students a case so that it develop his capacity to integrate information and resolve problems. To the start of the course will inform to the students of the procedure to be followed

Personalized assistance

Methodologies	Description
Case studies	They will offer tutorías personalised to guide the development of the cases posed

Assessment

	Description	Qualification	Training and Learning Results			
Objective questions exam	Subjects 1 to the 4. In the proofs will be evaluated the fundamental contents of the matter (masterclasses and practical) through objective questions (type test and short answer).	25	A1 A2	B1 B2 B6	C3 C16	D5
Objective questions exam	Subjects 5 to the 7. In the proofs will be evaluated the fundamental contents of the matter (masterclasses and practical) through objective questions (type test and short answer).	25	A1 A2	B1 B2 B6	C3 C16	D5
Case studies	The students will have to present the resolution of the case presented attending to the scale that will facilitate them to principle of course	20	A1 A2	B1 B2 B6	C5 C11 C12 C17	D5
Report of practices, practicum and external practices	The capacities and skills purchased during practise them will be evaluated of continuous form. The methodology of evaluation and weighting in the final note includes: 1- Implication of the student in the development of the practices. It will suppose 10% of the final qualification. 2- Delivery of reports of practices of laboratory. The reports will be made by each one of the subgroups of students organised in each group of practices. The half note obtained inform us will suppose 15% of the final note.	25	A1 A2	B1 B2 B6	C3 C11 C12	D5
Presentation	The students owe to present of oral way to resolution of the case chosen	5	A1 A2	B1 B2 B6	C11 C12 C16 C17	D5

Other comments on the Evaluation

Important: Independently that&*nbsp; the/the student/to choose CONTINUOUS Or GLOBAL EVALUATION the assistance to all the PRACTICES OF LABORATORY is COMPULSORY to APPROVE the matter (except the properly justified absences).The detection of plagiarism in the activities that make will suppose a qualification of 0 in the activity affected.Continuous evaluation:

1) Two partial proofs: each one will suppose 25% of the note. TO SURPASS The MATTER demands : the) a minimum of 4 points
(on 10) in each proof and *b) obtain a minimum half note of 5, calculated from the note obtained in the two partial.

2) Practical of laboratory: Implication of the student (10% of the final note) + practical reports (15% of the final note).

3) Seminar /study of cases: 20% of the final note+ presentation 5%.

To surpass the matter to sum: half note of the partial + practical note + notices case has to be equal or upper to 5.

The activities (partial proof, practical and cases) surpassed at the earliest opportunity of a course conserve for the second opportunity. In the second opportunity of a course can not recover practical and seminars, only can make the partial examinations no surpassed at the earliest opportunity.

To the students/the *repetidores/ace will conserve them the note of the practices and the seminars. Will have right to repeat the said
activities as long as they renounce by writing to the qualification obtained previously (document signed and envoy to the coordinator/the). The renunciation has to be done before they begin the practices.Global evaluation: The/the student/to that it choose global evaluation will have to surpass a final proof *integradora in which it will evaluate of the contents of the classrooms *magistrales, practices of laboratory and studies of case. The proof will consist in questions type test, short questions and resolution of problems/marry clinical.

To surpass the matter to note of the global proof will have to be equal or upper to 5. Of not to surpass the final proof, the qualification
of the/to student/to ONLY will be the obtained in the final proof *integradora on 10 points.

In the second opportunity of the course, the/to student/to suspense/to will have to be again evaluated of all the activities by means of a global proof.

If it does not surpass the matter in any of the opportunities of the course. The/to student/to will not have to do the practices, but yes
will be evaluated/to again of all the contents (classrooms *expositivas, practical and seminars), already was by means of continuous

or global evaluation.
General information
The academic calendar *pode consult in the following link:
http://bioloxia.uvigo.es/gl/docencia/horarios
The calendar of examinations *pode consult in the following link:
http://bioloxia.uvigo.es/gl/docencia/exámenes

Sources of information

Basic Bibliography

Emery; Turnpenny, **Elementos de genética médica y genómica**, 16, Elsevier, 2022

Dr. Álvaro González Hernández, **Principios de bioquímica clínica y patología molecular /**, 3, Elsevier, 2019

William B. Coleman, Gregory J. Tsongalis, **Molecular pathology: the molecular basis of human disease**, 2, Academic Press, 2018

Strachan T., Read A., **Human Molecular Genetics**, 5, Garland Science, 2018

Arsham M.S., Barch M.J., Lawce H.J, **The AGT Cytogenetics Laboratory Manual**, 4, Wiley-Blackwell, 2017

Complementary Bibliography

Recommendations

Subjects that are recommended to be taken simultaneously

Integrative cell biology and physiology: Implications for health/V02G031V01407
Clinical biochemistry and inmunology/V02G031V01405
Public health microbiology and parasitology/V02G031V01406

Subjects that it is recommended to have taken before

Biochemistry I/V02G031V01201
Biochemistry II/V02G031V01206
Genetics I/V02G031V01209
Genetics II/V02G031V01304
Technics in cellular and molecular biology/V02G031V01310