

Degree	Type	Year	Semester
4313802 Advanced Genetics	OT	0	1

Contact

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Teachers

Massimo Bogliolo

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Use of languages

Principal working language: english (eng)

Prerequisites

There are not specific requirement for this module. A certain background on human genetics and DNA damage response, including DNA repair mechanism, is advisable.

Objectives and Contextualisation

The aim of this module is to dissect the genetic basis of cancer predisposition. We will discuss and perform laboratory experiments to explore the molecular basis of syndromes such as hereditary breast cancer, familiar colon cancer or rare DNA repair syndromes characterized by cancer predisposition such as Xeroderma pigmentosum, Fanconi anemia or Lynch syndromes. We will also discuss and experimentally check how DNA repair can be targeted for specific anticancer therapies.

Skills

- Analyse the research results to obtain new products or processes valuing their industrial and commercial viability for transfer to society.
- Demonstrate a mastery of genetic analysis as a transversal tool applicable to any field of genetics.
- Demonstrate responsibility in management of information and knowledge.
- Design and apply scientific methodology in resolving problems.
- Develop critical reasoning in the area of study and in relation to the scientific and business environments.
- Integrate knowledge of the possible alterations in DNA with their consequences for living beings.
- Possess and understand knowledge that provides a basis or opportunity for originality in the development and/or application of ideas, often in a research context
- Student should possess an ability to learn that enables them to continue studying in a manner which is largely self-supervised or independent
- Students should be capable of integrating knowledge and facing the complexity of making judgements using information that may be incomplete or limited, including reflections on the social and ethical responsibilities linked to that knowledge and those judgements
- Students should know how to apply the knowledge they acquire and be capable of solving problems in new or little-known areas within broader contexts (or multidisciplinary contexts) related to their area of study

- Use and manage bibliographical information and other resources related to genetics and related fields.
- Use scientific terminology to argue the results of the research and show how to communicate in spoken and written English in an international setting.

Learning outcomes

1. Analyse the molecular mechanisms operating in genetic pathologies.
2. Analyse the molecular mechanisms operating in tumour processes and in the genetic predisposition to suffer them.
3. Analyse the research results to obtain new products or processes valuing their industrial and commercial viability for transfer to society.
4. Demonstrate responsibility in the management of information and knowledge and in the direction of groups and/or projects in multidisciplinary teams.
5. Develop critical reasoning in the area of study and in relation to the scientific and business environment.
6. Preparation and presentation of seminars
7. Preparation of work related to the module content
8. Students should be capable of integrating knowledge and facing the complexity of making judgements using information that may be incomplete or limited, including reflections on the social and ethical responsibilities linked to that knowledge and those judgements
9. Use and manage bibliographical information and other resources related to genetics and related fields.
10. Use scientific terminology to argue the results of the research and show how to communicate in spoken and written English in an international setting.
11. Write a report that considers the use of the methodology used in the module to resolve a specific problem
12. Write critical summaries about the taught seminars
13. Write reports on the genetic bases of different illnesses

Content

Theoretical lectures and seminars:

- Molecular basis of cancer predisposition: DNA repair and DNA damage response mechanisms.
- BRCAness and genetic predisposition to breast/ovarian cancer
- Rare DNA repair syndromes of cancer predisposition
- Targeting DNA repair in cancer treatment: chemosensitizing tumors by inhibiting DNA repair.

Synthetic lethality: Chemotherapy of BRCA tumours with PARP inhibitors

Laboratory experiments:

- Sensitivity of cells from Xeroderma pigmentosum to UV light
- Repair kinetics of UV-light induced pyrimidine dimers
- Sensitivity of BRCA negative cells to PARP inhibitors
- Homologous recombination assay

These experiments will require the following techniques:

- culturing of human cells from cancer predisposition syndromes
- cytotoxicity and cell cycle assays
- subnuclear local irradiation of fibroblast

- Plasmid transfection
- RNA interference by siRNA
- flow cytometry
- Western blot
- flow cytometry

Methodology

The student will receive several lectures to get the theoretical background required to perform and follow the laboratory experiments that will take most of the time in close contact with specialists in the field of this module. Bibliography will be given in advance via Campus Virtual so that the students can discuss it, summarize it or prepare PPT files in the format of seminars. The students will have to summarize the laboratory experiments performed in the laboratory.

Activities

Title	Hours	ECTS	Learning outcomes
Type: Directed			
Lectures	10	0.4	1, 2, 5, 6, 8, 9, 10, 12
Seminars	3	0.12	5, 6, 8, 10
Type: Supervised			
Discussion of results and preparation of PPT files	20	0.8	6, 12
Laboratory experiments	40	1.6	1, 2, 3, 7, 8, 10
Laboratory report	30	1.2	1, 2, 3, 4, 5, 7, 8, 9, 10, 11, 12, 13
Type: Autonomous			
Studing and reading	37	1.48	3, 4, 6, 9, 12

Evaluation

Students must pass a final exam to demonstrate that they have reached an acceptable degree of knowledge on the different topics discussed in the module.

Students will have to write a report of the experiments performed which will be corrected and evaluated.

Active participation in the laboratory and via Campus Virtual will be another source of information to reach the final qualification.

Evaluation activities

Title	Weighting	Hours	ECTS	Learning outcomes
Exam	40	2	0.08	1, 5, 8, 9, 10

Participation	20	8	0.32	1, 2, 3, 4, 5, 7, 9, 10, 11, 12, 13
Report	40	0	0	1, 6, 7, 10

Bibliography

The student will receive a list of references and the corresponding PDF documents via Campus Virtual