

**MOLECULAR CYTOGENICS IN ONCOLOGY**

1.- General information					
Code	303005	Plan		ECTS	3
Type	Elective	Course	2026/2027	Periodicity	2 <sup>st</sup> Semester
Language	Spanish				
Department	<a href="#">Cancer Research Center</a>				
Virtual Platform	<a href="https://cicloud.dep.usal.es/">https://cicloud.dep.usal.es/</a>				

1.1.- Faculty			
Professor Coordinator	Dr. Jesús M. Hernández Rivas		
Department	Medicine		
Research area	Haematology		
Center	<a href="#">Cancer Research Center</a>		
Office	Laboratory 12		
Tutorials	To be arranged		
URL Web	<a href="https://www.cicancer.org/grupo?id=31">https://www.cicancer.org/grupo?id=31</a>		
E-mail	<a href="mailto:jrivas@usal.es">jrivas@usal.es</a>	Phone	+34 923294819

Professor	Dra. Paola S. Dal Cin.		
Department	Harvard Medical School		
Research area	Pathology		
Center	<a href="https://connects.catalyst.harvard.edu/Profiles/display/Person/67560">https://connects.catalyst.harvard.edu/Profiles/display/Person/67560</a>		
E-mail	<a href="mailto:pdalcin@bwh.harvard.edu">pdalcin@bwh.harvard.edu</a>	Phone	

Professor	Dra. M Rocío Benito Sánchez Dr. Juan Luis García Hernández Dra. Norma Gutierrez Dra. Ana E Rodriguez Vicente Dra. María Abaigar Alvarado Dra Mónica del Rey Dr. Alberto Hernández Sánchez		
Research area	Haematology		
Center	Cancer Research Center Hospital,		
Office	Laboratory 12		
E-mail	<a href="mailto:beniroc@usal.es">beniroc@usal.es</a>	Phone	+34 923294812

	<a href="mailto:jl Garcia@usal.es">jl Garcia@usal.es</a> <a href="mailto:normagu@usal.es">normagu@usal.es</a> <a href="mailto:anaerv@hotmail.com">anaerv@hotmail.com</a> <a href="mailto:mymary@usal.es">mymary@usal.es</a> <a href="mailto:mdelrey@usal.es">mdelrey@usal.es</a>		
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## 2.- Previous recommendations

The student must attend the theoretical sessions having previously read and understood both the recommended bibliography, and the presentations that will be explained later. The first session will be focused on the sessions organization. Doubts and students comments will be discussed during this opening session. In the following sessions, the programme described below will be developed.

## 3.- Aims of the subject

To understand: 1. Genes and genetic alterations involved in the cancer development. 2. Epigenetic mechanisms involved in cancer development. 3. RNA mutations in tumor cells involved in cancer initiation, development and dissemination. 4. Genetic alterations study models (genomic editing, next generation sequencing and in vivo models).

To Know: 1. Methodologies frequently used in molecular cytogenetics: conventional cytogenetics, FISH, multicolored FISH, DNA arrays, expression arrays, next generation sequencing: NGS (genome, exome, transcriptome directed sequencing) and OGM (Optical Genome Mapping). 2. Cytogenetic alterations involved in cancer: numerical (gains and losses), structural alterations (translocation, inversion). 3. Genes undergoing methylation or acetylation in different types of cancer. 4. That genome and transcriptome analysis identifies cellular processes responsible for cancer development and metastases. 5. The role of molecular alterations in cancer diagnosis. 6. Limitations of these methodologies, identifying which of them are used at a diagnostic level and which are in an experimental phase. 7. The role of cancer of genome and transcriptome alterations in patients prognosis. 8. Genomic editing (CRISPR / Cas9) in the study and treatment of genetic alterations. 9. The main in vivo models in the study of molecular alterations. 10 Bioethical aspects.

## 4.- Skills to be acquired / Learning outcomes

### Skills

#### 4.1: Basic skills:

Acquire an exhaustive view of the classic and modern cytogenetic techniques applied to the diagnosis, prognosis and study of tumor molecular alterations.

#### 4.2: Specific skills:

- Recognize the tools for genomic and transcriptomic analysis of cancer.
- To Know how a cytogenetic study, a study of FISH, biochips, NGS and a basic bioinformatic analysis of samples of patients with leukemia or lymphomas, are done.
- To interpret a FISH study and an analysis of NGS data including, filtering, interpretation and classification of variants.
- To Know how a genome editing study by CRISPR is carried out in a leukemia model. To understand the implications of genome editing both as a study tool and as a therapeutic tool.

**4.3: Transferable skills:**

Team work. Results presentation. Discussion in small groups.

**5.- Contents (Syllabus)**

**Program:**

**Theoretical classes:**

**Block 1. Methodology**

Unit 1. Introduction to molecular cytogenetics in oncohematology. History, main methodologies. Conventional techniques in molecular cytogenetics: chromosomal study and fluorescence "in situ" hybridization (FISH). Multicolored FISH and comparative genomic hybridization.

Unit 2. Methodologies of genome analysis: microarrays and massive sequencing NGS.

Unit 3. Introduction to Sequencing. Classic sequencing methods. Next Generation Sequencing (DNA and RNA-seq)

Unit 4. The cytogenetic study in cancer diagnosis and prognosis. Main applications

Unit 5. NGS and OGM applications in hematology and Oncology.

**Block 2. Clinical Aspects**

Unit 6. Cytogenetic and molecular analysis in the study of acute leukemias

Unit 7. Cytogenetic and molecular analysis in the study of chronic hemopathies

Unit 8. Cytogenetic and molecular analysis of multiple myeloma.

Unit 9. Molecular cytogenetics of solid tumors. Soft tissue tumors.

Molecular alterations of sarcomas: classification. Ewing's tumor

Unit 10. Chromosomal study of solid tumors: problems. Molecular analysis of epithelial tumors.

Unit 11. Carcinomas: genomic studies.

Unit 12. Molecular analysis of other solid tumors: neuroblastoma, central nervous system tumors.

Unit 13. Pharmacogenetics and Pharmacogenomics in cancer. From genomic research to personalized therapy. Precision medicine. Pharmacogenes

**Block 3. Genome editing**

Unit 14. Animal models in the molecular study of cancer. Mouse models in the study of sarcomas.

Topic 15: Introduction to genome editing. Genome editing models: CRISPR. Generation of in vitro and in vivo models by CRISPR.

Topic 16: CRISPR applications in Oncology and hematology. Hematopoietic stem cell edition.

**Block 4. Genetics, cancer and bioethics**

Unit 17. Tumor heterogeneity and clonal evolution.

Unit 18. Ethical Implications of Personal Data Usage. Data protection regulation: research and clinical trials.

**Teaching Practices:**

1. Tumor cytogenetics: culture, collection and cell preparations. Staining, visualization and observation under the microscope.

2. Preparation and hybridization of specific fluorescent probes. Results analysis.
3. NGS sequencing. Data analysis.

**Seminars:**

Each group of students (2-3) will elaborate an oral presentation about one gene involved in cancer. For the seminar preparation, students should be documented with relevant scientific articles published recently. They can also make use of the documentation provided and discussed in the previous sessions. Presentations will be made and discussed with the participation of all students enrolled in the course.

**6.- Teaching methodology**

The student must attend the assessable theoretical sessions having previously read and understood both the recommended bibliography, and the presentations that will be explained later. The first session will be focused on the sessions organization. Doubts and students comments will be discussed during this opening session. In the following sessions, the programme described in the corresponding section will be developed.

Organization of the students in working groups that will consist of 2-3 students per group and that should prepare a class of those sessions included in the agenda. The selected classes will be adapted to the characteristics of the students to facilitate their involvement in the study.

The student must attend the seminars (6 hours) in which each group will present a published research paper on any of the topics discussed in the course and a critical evaluable dialogue will be established.

Two clinical cases will be showed to the students. Students should resolve these cases by using the contents showed in the sessions. They can make use of the bibliographic sources available, especially the internet addresses provided in the theoretical sessions.

**6.1.- Estimated learning time**

	Hours tutored by the teacher		Individual work (hours)	TOTAL HOURS
	Attendance required (hours)	Distance learning (hours)		
<b>Lectures</b>	20			20
<b>Practices</b>	- In classroom			
	- In laboratory		8	8
	- In computer classroom			
	- Countryside			
	- Others (specify)			
<b>Seminars</b>				
<b>Work presentations and debates</b>	10			10
<b>Tutorials</b>	2			2
<b>Online activities</b>	5	5		10
<b>Work preparation</b>		5	5	10
<b>Other activities</b>		5		5
<b>Exams - evaluation</b>	2	8		10
<b>TOTAL</b>	<b>39</b>	<b>23</b>	<b>13</b>	<b>75</b>

### 7.- Materials, other bibliographical, electronic references or any other type of resource

Cancer Cytogenetics: Chromosomal and Molecular Genetic Aberrations of Tumor Cells, Fourth Edition. Editor(s): Sverre Heim and Felix Mitelman. 26 June 2015. Wiley Blackwell  
Print ISBN:9781118795538 |Online ISBN:9781118795569 |DOI:10.1002/9781118795569.

1. Atlas of Genetics and Cytogenetics in Oncology and Haematology.

<http://atlasgeneticsoncology.org/>

2. GeneCards®: The Human Gene Database. <https://www.genecards.org/>

3. PharmGKB. <https://www.pharmgkb.org/>

4. Wellcome Sanger Institute. <https://www.sanger.ac.uk/>

### 8.- Assessment

#### 8.1: Assessment Criteria:

#### 8.2: Assessment Systems:

Class attendance is mandatory to be evaluated.

Participation in theoretical sessions and debates (50% of the final grade).

Evaluation of the course in writing (50% of the final grade).

#### 8.3: General Considerations and Recommendations for Assessment and Resit:

### 9.- Weekly Teaching Schedule