

**Molecular Histopathology: from Laboratory to Clinic**

Code: 103638  
ECTS Credits: 3

Degree	Type	Year	Semester
2502442 Medicine	OT	3	0
2502442 Medicine	OT	4	0
2502442 Medicine	OT	5	0
2502442 Medicine	OT	6	0

### Contact

Name: Francisco Javier Andreu Navarro

Email: FranciscoJavier.Andreu@uab.cat

### Use of Languages

Principal working language: catalan (cat)

Some groups entirely in English: No

Some groups entirely in Catalan: Yes

Some groups entirely in Spanish: No

### Teachers

Carmen Blazquez Maña

Inmaculada Méndez Coca

Rubén Carrera Salas

### Prerequisites

It is necessary for the student to assimilate basic skills in Biochemistry and Molecular Biology and also in Physiology and Pathology.

### Objectives and Contextualisation

The subject aims to introduce the student to the clinical applications of Molecular Pathology, both from the point of view of diagnosis, and in the detection of molecular alterations of predictive interest in personalized oncological treatments.

### Competences

Medicine

- Demonstrate an understanding of the fundamentals of action, indications, efficacy and benefit-risk ratio of therapeutic interventions based on the available scientific evidence.
- Demonstrate basic research skills.
- Demonstrate understanding of the basic sciences and the principles underpinning them.
- Demonstrate understanding of the mechanisms of alterations to the structure and function of the systems of the organism in illness.

- Demonstrate understanding of the organisation and functions of the genome, the mechanisms of transmission and expression of genetic information and the molecular and cellular bases of genetic analysis.
- Indicate the basic diagnosis techniques and procedures and analyse and interpret the results so as to better pinpoint the nature of the problems.
- Write patient records and other medical documents that can be understood by third parties.

## Learning Outcomes

1. Analyse information from biological sequencing.
2. Correctly write reports on the results of different types of tests (analytic, genetic).
3. Demonstrate basic research skills.
4. Describe the diagnosis, prognosis, prevention and treatment for the most common genetic pathologies in the human population.
5. Describe the indications of anatomopathological tests.
6. Describe the molecular basis of the mechanisms underlying anatomopathological alterations of various diseases, primarily neoplastic and hereditary ones, in different body systems.
7. Identify the concept of medical bioinformatics and the integration of genetic and clinical databases.
8. Identify the most efficient tests for prevention, diagnosis and control of treatment for the most common human pathologies.
9. Relate genetic dysfunction to the pathological phenotype.

## Content

Specialized seminars

Topic 1

Applied molecular biology (introduction to the primary structure of nucleic acids, superior structures and condensation of DNA, cell cycle and organization of the eukaryotic genome, replication and transcription, genetic code and mechanisms of regulation of translation).

Topic 2

Molecular basis of mutations and DNA repair (concepts, classification of mutations and diseases associated with alterations of repair).

Theme 3

Diagnostic molecular pathology in lymphoid neoplasms (translocations and clonality studies). Applications of molecular techniques in the diagnosis of solid tumors

Theme 4

Hereditary genetic factors and diagnostic molecular pathology.

Topic 5

Mutational study in cancer and new therapeutic targets.

Laboratory practices

Seminar 1

Nucleic acids extraction: DNAs and RNAs. Protein extractions. Performance of different types of samples (frozen tissue, biopsies, cell blocks, cytologies). Different types of extraction. Other basic techniques: quantification by spectrophotometry, fluorometry. Electrophoresis techniques (agarose, acrylamide). RNA quality (RIN) and protein quality (DIGE).

## Seminar 2

PC Techniques: Bases. Type (Multiplex-PCR, AS-PCR, Nested-PCR, COLD-PCR, RT-PCR (TaqMan, FRET, ...). Osna. Sequencing Sanger, pyrosequencing Conventional cytogenetic techniques: FISH (type of probes, CEP / LSI / WCP, hybridization strategies: Dual-Color Break Apart, Dual-Color Dual-Fusion, amplification probes). CISH. SISH. CGH.

## Seminar 3

Techniques applicable to hematopathology: Study of clonality in B and T lymphomas (BIOMED-2). Studies of specific translocations (MYC, BCL-2, CCND1). Studies of translocations in solid tumors (EWSR1, SYT).

## Seminar 4

Screening of Lynch Syndrome. Molecular techniques PCR and IHC. Description, results, discordant cases, results, genetic counseling.

## Seminar 5

Pyrosequencing technique (alternatives: RT-PCR, ARMS-PCR, HRM, Sanger). Advantages and limitations. Approach from different types of samples (study yield) according to the type of neoplasia.

## Methodology

For the 2019-2020 academic year, the professor designated by the Department as responsible for the subject at the Faculty level is:

UDPT

Francisco Javier Andreu  
xandreu@tauli.cat  
(12 students)

The students, divided into groups of 3 or 4 students, will face real clinical problems that they will have to develop from the initial diagnostic material (paraffin block), through the DNA extraction and quantification method, PCR / hybridization, and sequencing , until the issuance of the molecular pathology report.

The selected cases of the pathology file may include mutational study of lung and colon cancer, study of a family with suspected HNPCC (Lynch syndrome) or diagnosis of limfoproliferatiu process, among others.

## Activities

Title	Hours	ECTS	Learning Outcomes
Type: Directed			
Laboratory practices (PLAB)	7.5	0.3	
Specialized Seminars (SESP)	7.5	0.3	
Type: Supervised			
Assistencial practicum without guidelines	7.5	0.3	
Virtuals	7.5	0.3	
Type: Autonomous			

Personal study, bibliography consultation, problem solving, work accomplishment	41	1.64	1, 3, 4, 6, 5, 7, 8, 2
---------------------------------------------------------------------------------	----	------	------------------------

## Assessment

The marks for the final qualification of the subject will be:

- The resolution of problems in class (20% of the final grade)
- Oral presentation (50% of the final grade). Assessment will take into account the qualification of the presentation and the attitude in class. The non-active participation in the presentation of the corresponding presentation will prevent the qualification of this part of the subject.
- The evaluation of the work (30% of the final grade). The student will have to make a presentation on the case that will have developed during the intensive practice.

Students who fail to carry out both theoretical and practical evaluation tests will be considered as Not evaluated by exhausting the rights to the matriculation of the subject.

Recovery system:

For students who have not passed the subject through the scheduled assessment activities, they can participate in an oral recovery test, according to the programmed exam calendar.

## Assessment Activities

Title	Weighting	Hours	ECTS	Learning Outcomes
Delivery of work / reports	30%	1	0.04	1, 3, 4, 6, 5, 7
Evaluation through practical cases and problem solving in class	20%	1	0.04	6, 5, 8, 2, 9
Oral defense of works	50%	2	0.08	6, 5

## Bibliography

Specific bibliography

Molecular biology and genetic engineering. Luque J, Herráez A. Ed. Elsevier Science, Madrid, 2002.

Lliçons de Molecular Pathology. González Sastre F, Guinovart JJ. Ed. Springer-Verlag Ibérica, Barcelona, 2000.

Molecular Biology of the Gene (2006) Watson et al. Editorial Panamericana.

Reference bibliography

Molecular Biology of the Cell. Alberts B. Ed. Omega, Barcelona 2010.

Internet resources

<http://www.sanger.ac.uk/genetics/CGP/cosmic/>

[www.ncbi.nlm.nih.gov/pubmed](http://www.ncbi.nlm.nih.gov/pubmed)

[www.ncbi.nlm.nih.gov/omim](http://www.ncbi.nlm.nih.gov/omim)

<http://www.hgvs.org/mutnomen/>