

## **COURSE DATA**

Data Subject		
Code	34473	
Name	Oncology and medical genetics	
Cycle	Grade	
ECTS Credits	6.0	
Academic year	2022 - 2023	

Study (s)
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Degree Center Acad. Period

year

1204 - Degree in Medicine Faculty of Medicine and Odontology 4 Second term

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### **Subject-matter**

Degree	Subject-matter	Character
1204 - Degree in Medicine	13 - Human clinical training II	Obligatory

### Coordination

Name

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Department

260 - Medicine

285 - Pathology

## SUMMARY

Half of the subject will develop the topics about Oncology and the other half of the subject will develop the topics of Medical Genetics.

Oncology is the medical specialty that deals with the analysis and treatment of malignant tumours. Oncology, therefore, is in charge of the detection, treatment and control of neoplasm diseases. In the case of treatment, it considers the possibility of subjecting the patient surgery and non-surgical therapies, such as chemotherapy, radiotherapy and molecular treatments. On the other hand, Oncology seeks to provide palliative care to all who suffer terminal diseases and approaches focalized genetic tests focused on the detection of tumours. The general objectives are to train professionals with theoretical and practical knowledge, attitudes and skills that enable the student to solve and guide the medical situations that condition tumour disease.



Human Genetics is the scientific discipline that studies the human species heritage. The Medical Genetics is the medical-sanitary specialty that applies the knowledge of this discipline to the medical practice, dealing with the diseases of genetic origin, including hereditary and malformed or disabling pathologies of the human species. It includes all the aspects related with the aforementioned: etiology, physiopathology, heritage mechanisms, population screening, diagnosis (clinical and laboratory), prognosis and recurrence risk (genetic counselling), treatment and prevention, including prenatal and postnatal stage of the person.

## PREVIOUS KNOWLEDGE

### Relationship to other subjects of the same degree

There are no specified enrollment restrictions with other subjects of the curriculum.

### Other requirements

## **OUTCOMES**

### 1204 - Degree in Medicine

- Obtain and elaborate a clinical history withrelevant information.
- Perform a physical examination and a mental health assessment.
- Have the capacity to make an initial diagnosis and establish a reasonable strategy of diagnosis.
- Establish the diagnosis, prognosis and treatment, applying principles based on the bestinformation available and on conditions of clinical safety.
- Indicate the most accurate therapy in acute and chronic processes prevailing, as well as for terminally ill patients.
- Plan and propose appropriate preventive measures for each clinical situation.
- Acquire properclinical experience in hospitals, health care centres and other health institutions, under supervision, as well as basic knowledge of clinical management focused on the patient and the correct use of tests, medicines and other resources available in the health care system.
- Know how to use the sources of clinical and biomedical information available, and value them critically in order to obtain, organise, interpret and communicate scientific and sanitary information.
- Know how to use IT in clinical, therapeutic and preventive activities, and those of research.
- Proper organisation and planning of the workload and timing in professional activities.
- Team-working skills and engaging with other people in the same line of work or different.
- Criticism and self-criticism skills.



- Capacity for communicating with professional circles from other domains.
- Acknowledge diversity and multiculturality.
- Consideration of ethics as a fundamental value in the professional practise.
- Working capacity to function in an international context.
- Is aware of tumour disease, its diagnose and management.
- Knows how to make a diagnosis and apply genetic counselling.
- Knows how to perform a complete anamnesis, focused on the patient and orientated to various pathologies, interpreting its meaning.
- Knows how to evaluate modifications in clinical parameters at different ages.

## **LEARNING OUTCOMES**

- Learn about the main clinical profiles related with the oncological pathology.
- Learn how to perform an anamnesis and complete exploration, focused on the patient and oriented towards Oncology, interpreting its meaning.
- To elaborate a diagnostis and to establish a therapeutic strategy.
- The student will be able to design a follow-up plan about the patient and his/her disease.
- Knowledge of the Genetic theoretical bases.
- Learn the hereditary nature of the diseases and their mechanism of transmission.
- To learn the prevalence and epidemiology of genetic diseases in general population or, in its case, in a determined group of population.
- To know and to apply the bases and concepts of basic genetics, human genetics, medical genetics and population genetics to the calculation of recurrence risk in hereditary diseases, including the possible modifier factors of the same one. Apply this knowledge to the practice of clinical genetics.
- To know the theoretical bases of genetic counselling, including the calculation of recurrence risk of the hereditary diseases.
- To be familiar and use in a practical and effective way the text books, scientific journals, computer databases and other sources of information as useful tools in the practice of medical genetics in each of its areas.

## **DESCRIPTION OF CONTENTS**



## 1. THEORETICAL LESSONS: Oncology

- 1. Cancer epidemiology: incidence, prevalence and mortality. Tumours registry. Etiologic factors. Heritage predisposition. Cancer pathogenesis.
- 2. Diagnosis and natural history of cancer. Primary and secondary prevention. Cancer clinical manifestations. Diagnosis, extension diagnosis and prognosis factors.
- 3. Cancer multidisciplinary approach. The tumours committees. Treatment principles of cancer. Medical treatment of cancer: chemotherapy and hormonal therapy.
- 4. Biological treatment of cancer. Guided therapies against molecular targets. Immunotherapy.
- 5. Research in Oncology. Clinical essays.
- 6. Palliative and support treatment. Control of symptoms in patients with advanced tumours. Treatment toxicity: prevention and control strategies.
- 7. Germinal tumours with a model of curable neoplasm with chemotherapy.
- 8. Breast cancer as a model of treatable neoplasm according to its molecular profile.
- 9. Colon and rectal cancer as a model of treatable neoplasm in a multidisciplinary approach.
- 10. Lung cancer as a model of heterogeneous neoplasm with different treatment profiles according to their molecular profile.
- 11. Ovarian cancer as a model of the necessity of therapeutic innovation.
- 12. Prostate cancer.

#### 2. THEORETICAL LESSONS: Medical Genetics

- 1. Introduction. Basic principles in genetics. Role of the genetics in medicine. Types of genetic diseases. Clinical impact of the genetic disease.
- 2. Monogenetic diseases: detection of genetic diseases in medical practice.
- 3. Monogenetic diseases. Modifier factors. New mutation. Mosaicism of the germinal line. Delay in the age of presentation: example, Huntington disease. Reduced penetrance: example, retinoblastoma heritage.
- 4. Monogenetic diseases: modifier factors (II). Variable expressivity: example, Neurofibromatosis. Pleiotropy and locus heterogeneity: example, Marfan's syndrome. Genomic imprinting, epigenetic changes: example, Prader-Willi and Angelman's syndromes. Uniparental disomy.



- 5. Diseases linked with the X chromosome, transmission patterns. Lethal alleles in homozygous. Most frequent recessive diseases. Inactivation of the X chromosome and genes expressions linked with the X chromosome. Genetics of the Fragile X chromosome Syndrome (FXS). Limited phenotypes or influenced by gender.
- 6. Principles of the multifactor disease. Multifactor disorders. Principles of the multifactor disease. Search of candidate genes. Genetic and environmental base. Load/threshold model. Most frequent diseases that affect the adult population: cardiovascular diseases, hypertension, familial hypercholesterolemia, etc.

### 3. THEORETICAL LESSONS: Medical Genetics (continous)

- 7. Clinical cytogenetic. Chromosome anomalies detection. Autosomal disorder. Incidence. Clinical indications for the chromosomal analysis. Trisomies and viable monosomy. Risk factors. Chromosomal anomalies and clinical phenotypes: trisomy 21, trisomy 13, trisomy 18, etc.
- 8. Clinical cytogenetics (II). Gonosomopathies. Chromosomal basis of sexual determination. Aneuploidies of the sex chromosomes in women: Turner syndrome, chromosomal variants, trisomy X. Aneuploidies of sex chromosomes in men: Klinefelter syndrome, double Y syndrome, structural abnormalities of the Y chromosome. Genetic bases of sexual differentiation. XX men. XY women.
- 9. Genetic bases of cancer. Genes involved: protooncogenes and tumor suppressor genes. Genetic changes involved in tumor progression. Inheritance of cancer susceptibility.
- 10. Population genetics. Genetic variation in human population. Genetic structure of the human population. Gene frequencies. Hardy-Weinberg's equilibrium. Genetic drift, migration and gene flow. Evolution.
- 11. Genetic advise and risk evaluation. Indications for the genetic advise. Prenatal diagnosis. Methodology.

### 4. PRACTICAL LESSONS: Oncology

- 2. Seminar practices:
- Discussion of cases of breast cancer
- Discussion of cases of lung cancer
- Discussion of cases of colon cancer
- Oncological emergencies
- Oncological support treatment: cancer pain/myelosuppression
- 3. Clinical practices



### 5. PRACTICAL LESSONS: Medical genetics

Problem solving

- 1. Mutations analysis. Molecular diagnosis. Databases management.
- 2. Resolution and interpretation of genealogical trees. Problem solving of Mendelian genetics.
- 3. Analysis of structural genetic variations. Databases management.
- 4. Cytogenetic problem solving.

#### **Seminar Practices**

- 1.- Molecular diagnosis techniques
- 3.- Genetic bases of the metabolism diseases.
- 4.- Epigenome and disease. MicroRNAs, molecular bases and applications.
- 5.- Patrón de transmisión de las enfermedades mitocondriales. Miopatías mitocondriales.
- 6.- Presentation of clinical cases: monogenetic and chromosomal.

## WORKLOAD

ACTIVITY	Hours	% To be attended
Seminars	26,00	100
Theory classes	26,00	100
Clinical practice	23,01	100
Development of individual work	15,00	0
Study and independent work	40,00	0
Preparation of practical classes and problem	20,00	// / plb 0
TOTAL	150,01	

## **TEACHING METHODOLOGY**

In the **theoretical lessons**, the teacher will expose, through a master class, the most important concepts and contents in a structured way, to obtain of the knowledge and skills that the students must acquire. The students' participation will be encouraged. The teaching materials used by the professor will be available, if he considers it appropriate, through the electronic resource Aula Virtual.

Classroom practices: **seminars**. In small groups, the teacher will set specialized topics in depth, case studies, bibliography handling, current topics... the group work and oral presentation will be encouraged. It can be understood as "cooperative learning".

**Clinical practices**: students' clinical practices in sanitary services in the different university hospitals, primary health centres, mental health centres, public health areas, in order to learn how to perform an anamnesis and basic clinical explorations, with a first contact with patients, supervised by the professor.



# **EVALUATION**

**Theoretical assessment**: 50% of the final mark. It will be done through a written test that will be about the content of the program and will have as the main objective to assess the knowledge acquisition. The content of the test will be the same for each group of the same subject.

**Practical assessment**: 50% of the final mark. It will be done by the assessment of the participation in the different activities and with the fulfilment of a test that assesses the acquisition of skills related with the general and specific competencies.

Regarding the **Oncology** part, the assessment will be as follows:

- Theory, 5 points: 30 multiple-choice questions, 5 options. Wrong answers will not subtract any point. Blank answers will be counted as 0
- Practice, 5 points: 30 multiple-choice questions. 5 options. Wrong answers will not subtract any point. Blank responses will be counted as 0

Regarding the **Medical Genetics** part, the assessment is as it follows:

- Teoria, 5 punts: 40 preguntes tipus test, 4 opcions cada una. Cada resposta errònia resta la tercera part d'una resposta encertada. A les preguntes no contestades se les donarà un valor de zero.
- Practices, 5 points: Informatics exercises: 0.5 points (0.25 each one). Exam: 4.5 points. It will contain several practical exercises and short seminar questions.

To achieve the final mark, the student must past both the Oncology part and the Medical genetics part. The marks of both areas in the theoretical and practical assessment are divided between 2.

In order to access to an advance on the call of this subject, it is a requirement that the student has coursed all his/her practices.

Attendance to practical sessions is mandatory. Unjustified non-attendance to more than 20% of the sessions will make it impossible to pass the course.

Students are reminded of the importance of carrying out evaluation surveys on all the teaching staff of the degree subjects.



## **REFERENCES**

#### **Basic**

- THOMPSON & THOMPSON. Genética en Medicina. Ed. Masson, 8ª edición.
- LYNN B. JORDE. Genética Médica. Ed. Elsevier 4ª edición.
- RAFAEL OLIVA VIRGILI. Genética Médica. Ed. Díaz de Santos.
- EMERY and RIMOIN'S. Principles and Practice of Medical Genetics. Ed. Elsevier, 6a edición.
- EMERY. Elementos de Genética Médica. 15ª edición
- HARRISONS PRINCIPLES OF INTERNAL MEDICINE. Ed McGraw-Hill Education. 20ª edición. 2018.
- DEVITA, HELLMAN AND ROSENBERG'S CANCER: PRINCIPLES AND PRACTICE OF ONCOLOGY. Wolters Kluwer Health Library. 11a edición. 2018.
- Recursos-e Salut: ClinicalKey Student. Elsevier (Scopus, ScienceDirect): uv-es.libguides.com/RecursosSalut/BibliotecaSalut

