Subject 7. STRUCTURAL HEMOGLOBINOPATHIES

Code:

Type: Optional

Schedule: To be defined

Departments involved: Medicine

Coordinator:
Joan-LLuis Vives Corrons (Departament de Medicina, Universitat de Barcelona, Unidad de Patología Eritrocitaria)

Academia:

1. Jacques Elion
2. Beatrice Gulbis
3. Lucia de Franceschi
4. Adlette Inati
5. Mariane de Montalembert
6. Frédéric Galactéros

Subject Coordinador: Béatrice Gulbis

Credits ECTS: 3

Subject total teaching time (in hours):75

- **Presential (teacher): 50**
- **Autonomous (student): 25**

Requirements for subject learning

Skills to be developed

TRANSVERSAL SKILLS

- Being able to interact with other medical specialists to advise them
- Ability to work in interdisciplinary teams and collaborate with other researchers together, act independently and use initiative
- Ability to teach and disseminate knowledge in the social environment in both expert and non-expert audiences, clearly and in different languages
- Ability to integrate knowledge and ways to deal with the complexity and formulate judgments based on limited information, but so thoughtful, considering social and ethical repercussions of the trials
- To keep up to date knowledge exposed in the field of the international scientific community, that is, to seek, obtain and interpret information obtained in biomedical databases and other sources
- Being able to know the principles of bioethics and medico-legal research and professional activities in the field of biomedicine

SPECIFIC SKILLS

- Knowing the epidemiological, pathogenic, clinical and therapeutic advances of major haemoglobinopathies due to structural defects.
- Know the most advanced and complementary clinical diagnostic examinations of major structural haemoglobinopathies and sickle-cell disease.
- Be able to recognize, interpret and diagnose properly laboratory abnormalities present in patients with haemoglobinopathies ,in general, and in cases of sickle-cell disease (SCD) .
- To develop, implement and evaluate clinical practice guidelines for patients with sickle-cell disease (SCD).

Subject Learning Objectives

A. General Objectives

The main objective of the course is to help train clinicians and researchers in the field of major haemoglobinopathies and SCD in a very well defined patient's multidisciplinary care unit with an area of excellence for translational research. Haemoglobinopathies and especially SCD represent a clinical social health problem with a high associated morbidity and increasing incidence due to immigration impact from Sub Saharan african populations.

B. Specific Objectives

To know in depth the erythropoiesis and its defects, their etiological mechanism/s and their main clinical manifestations and research possibilities. Furthermore, to assess the results of clinical trials in the international development of new biological treatments cost-effectiveness studies.

Teaching programme

Subject 6: Rare anaemias due to iron and vitamins metabolic disorders

Date	Topic	Chapter	Professor	Language
	7.1 General hemoglobin and hemoglobinopathies	7.1.1 Structure of Hemoglobin (4h)	Jacques Elion	English
		7.1.2 Evolution and geographical	Jacques Elion	English
		7.1.3 Genetics of hemoglobin and its molecular variant (4h)	Jacques Elion	English
		7.1.4 Classification of hemoglobinopathies (2h)	Jacques Elion	English
		7.1.5 Genetic counseling and prenatal diagnosis (2h)	Jacques Elion	English
	7.2 Sickle cell disease and related syndromes (26h)	7.2.1 Homozygous sickle cell anemia (Sickle Cell Syndrome) (3h)	Beatrice Gulbis	English
		7.2.2 Clinical and laboratory diagnosis (4h)	Beatrice Gulbis	English
		7.2.3 Investigations. Imaging techniques (4h)	Lucia de Franceschi	English
		7.2.4 Treatment and clinical follow (4h)	Lucia de Franceschi	English
		7.2.5 Neonatal screening (3h)	Adlette Inati	English
		7.2.6 Heterozygous form with sickle cell disease (2h)	Lucia de Franceschi	
		7.2.7 Clinical and laboratory diagnosis in children and adults (2h)	Lucia de Franceschi	English
		7.2.8 Molecular diagnosis (2h)	Jacques Elion	English
		7.2.9 Treatment and clinical follow (2h)	Mariane de Montalembert	English
	7.3 Structural hemoglobinopathies not due to HbS (8h)	7.3.1 Hemoglobin C (HbC) (2h)	Frédéric Galactéros	English
		7.3.2 Hemoglobin E (HBE) (2h)	Jacques Elion	English
		7.3.3 Hemoglobin D (HbD) (2h)	Jacques Elion	
		7.3.4 Unstable hemoglobinopathies (2h)	Frédéric Galactéros	English
	7.4 Association of HbS with other hemoglobinopathies or red blood cells defects (2h)		Lucia de Franceschi	English

Methodology and General Organisation

A. **Main Lectures**: They will have a duration of 60 minutes; The first 40 minutes will be devoted to the exhibition of the teaching topic by the teacher and the remaining 20 minutes will be devoted to the interaction between students and teacher on the key issues of teaching topic theme (18 lectures= 18 hours).

B. **Interactive Seminars**: Will last 60 minutes and they will present case studies that the approach to analyze diagnostic and therapeutic evolution of patients with major erythropoietic defects (5 seminars = 5 hours).

C. **Student supervised task**: Students will prepare for approximately 1 hour each of the teaching classes / seminars and, for this, the teacher will provide a minimum of 2 articles in PDF format on the topic of the corresponding subject (class or seminar) (25 x 1 hour classes / seminars = 25 hours).

D. **Self Assessment** : At the end of the course (maximum two weeks after the last lecture), students must submit a portfolio summarizing skills acquired in this course (Independent task = 25 hours).

Attendance and degree of participation in lectures and interactive seminars (40%)
Realization of autonomous work, presentation and discussion with the teacher (60%)

Essential information resources

RELEVANT BIBLIOGRAPHY