



UNIVERSITAT DE BARCELONA



Facultat de Medicina

Plan Docent de la Asignatura 3: “Erythrocyte metabolism defects”

Màster en Competències Mèdiques Avançades - Anemias Raras i síndromes relacionados

GENERAL DATA

Subject: 3. ERYTHROCYTE METABOLISM DEFECTS

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. Richard van Wijk
2. Joan-Lluis Vives Corrons
3. Maria del Mar Mañú
4. Adlette Inati
5. Mariane de Montalembert
6. David Rees
7. Paola Bianchi

Subject Coordinador: Paola Bianchi

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 RBC enzyme defects and metabolic abnormalities
- Know the most advanced and complementary clinical diagnostic tests for the diagnosis of RBC enzymopathies
- Be able to recognize, interpret and diagnose properly laboratory abnormalities of the erythroenzymopathies
- To develop, implement and evaluate clinical practice guidelines for patients haemolytic anaemia due to RBC defects.

Subject Learning Objectives

A. General Objectives

The main objective of the course is to help training clinicians and researchers in the field of quality major erythropoietic defects in a very well defined as a unit of care is an area of excellence for translational research. Erythropoietic defects represent a systemic problem with increasing incidence and high associated morbidity.

B. Specific Objectives

To know in depth the haemolytic anaemia (acute and chronic) due to RBC enzymopathies, its ethiological mechanism/s, main clinical manifestations and research possibilities. Furthermore, to assess the results of clinical trials in the international development of new biological and /or genetic therapy based on cost-effectiveness studies.

Teaching programme

Subject 3: “Erythrocyte metabolism defects”

Date	Topic	Chapter	Professor	Language
	3.1 Overview of metabolic enzymes (8h)	3.1.1 Red blood cell metabolism (3h)	Richard van Wijk	English
		3.1.2 Structure and genetics of RBC	Richard van Wijk	English
		3.1.3 Classification and function of RBC enzymes (2h)	Richard van Wijk	English
				English
	3.2 Glucose	3.2.1 Clinical and laboratory diagnosis	Joan-Lluis Vives Corrons	English
		3.2.2 Molecular diagnosis (3h)	Maria del Mar Mañú	English
		3.2.3 Treatment and clinical follow (2h)	Adlette Inati	English
	3.3 Deficit of pyruvate kinase (PK)	3.3.1 Clinical and laboratory diagnosis (2h)	Joan-Lluis Vives Corrons	English
		3.3.2 Molecular diagnosis (2h)	Maria del Mar Mañú	English
		3.3.3 Treatment and clinical follow (2h)	Mariane de Montalembert	English
	3.4 Enzymopathies glutathione metabolism (10h)	3.4.1 Gluthation and RBC antioxidant mechanisms (2h)	Richard van Wijk	
		3.4.2 GGS synthetase and glutathione synthetase (2h)	Richard van Wijk	English
		3.4.3 Glutathione reductase (GP) (2h)	Richard van Wijk	English
		3.4.4 Glutathione peroxidase (GP) (2h)	Richard van Wijk	English
		3.4.5 6 Phosphoglucosate dehydrogenase (6PGD) (2h)	Richard van Wijk	English
	3.5 Very rare glycolytic	3.5.1 Phosphoglucose isomerase deficiency (PGI) (2h)	Joan-Lluis Vives Corrons	English
		3.5.2 Hexokinase deficiency (HK) (2h)	Richard van Wijk	
		3.5.3 Triosephosphate deficiency (TPI) (2h)	David Rees	English
		3.5.4 Phosphofructokinase deficiency (PFK) (2h)	Paola Bianchi	English
		3.5.5 Phosphoglycerate kinase Deficiency (PGK) (2h)	Paola Bianchi	English

		3.5.6 Aldolase deficiency and other VR enzymopathies (2h)	Paola Bianchi	English
		3.5.7 Phosphoglucomutase (PGM) deficiency (2h)	David Rees	English
	3.6 Enzymopathies of nucleotide	3.6.1 Pyrimidine 5 'nucleotidase (P5'N) deficiency (2h)	David Rees	English
		3.6.2 Adenosine deaminase hyperactivity (ADA) (2h)	To be defined	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