Nom de l'assignatura: Molecular basis of sensory disorders

Codi: (a emplenar posteriorment)

Tipus: Optativa

Impartició: English

Departaments implicats: Ciències Fisiològiques II, Patología y Terapéutica Experimental

Nom del professor coordinador: Dra. Ana Méndez Zunzunegui

Membres de l'equip docent: Dra. Ana Méndez Zunzunegui, Dr. Jordi Llorens.

Crèdits ECTS: 3

Hores estimades de l'assignatura: 75
  • Hores presencials 30
  • Hores aprenentatge autònom 45

Prerequisits per cursar l'assignatura

Basic knowledge of English language for written and oral communication.

Competències que es desenvolupen en l'assignatura

- Capacity to form a historical perspective and current status of a scientific field from a thorough revision of the literature.
- Comprehend that significant advances in basic biomedical research can translate to amelioration of patients by using recent examples from the visual field.
- Capacity to present and critically discuss seminal articles in the field, as well as more recent breakthrough articles, clinical trials and basic research forefronts.

Objectius d'aprenentatge de l'assignatura

Main Aims

- Develop the capacity to comprehend the current status and challenges of a particular scientific field of biomedical research from a thorough revision of past and present literature.
- Acquire the ability to get a global vision of a field (e.g. sensory disorders) and grasp the importance of applying multidisciplinary approaches to make significant advances in the field.
- Comprehend that basic research should ultimately serve translational research.
Familiarize students with scientific communications and exchanges in English.

Specific Aims

- Promote active participation of students in the class, whatever their English level. Students will be expected to ask questions in lectures, and to actively participate in critical discussions of the papers presented in seminar sessions.

- Students will acquire a basic knowledge of sensory systems, their cellular and molecular biology, as well as how genetic defects in sensory systems lead to inherited sensory disorders. We will review the research fronts that are aimed at ameliorating or curing these disorders, such as the use of genetic diagnosis, gene therapy, trophic factor and anti-oxidant treatments, cell replacement and biomedical engineering devices.

**Bloc temàtic o de continguts de l’assignatura**

**LECTURE SCHEDULE:**

1. **Introduction to the organization of the sensory systems.** AMZ

2. **The chemical senses: taste and smell.** JLB

3. **The eye and the retina.** AMZ

4. **Inherited retinal diseases I.** AMZ
   Genetic heterogeneity of retinal inherited disorders. Mutations that primarily affect rod function: retinitis pigmentosases (RP) and congenital stationary night blindness (CSNB). Gene defects affecting primarily cone function: cone dystrophies. Severe congenital forms of blindness: Leber Congenital Amaurosis, LCA.

5. **Inherited retinal diseases II.** AMZ

6. **The central visual system: agnosias** AMZ

7. **The auditory system.** JLB

8. **Molecular basis of inherited deafness.** JLB
9. The vestibular system JLB
Transduction in the vestibular system.

10. Ciliopathies affecting sensory systems AMZ

11. The somatic sensory system I. JLB
12. The somatic sensory system II. JLB
13. Somatic sensory disorders. JLB

14. Mutation discovery and genetic testing of inherited sensory disorders. AMZ

15. The prospects of gene therapy and pluripotent stem cell therapies. AMZ
How gene therapy and pluripotent stem cell therapies are changing the prospects to cure inherited sensory disorders. Examples from the retina.

SEMINAR SCHEDULE:

1. The chemical senses: taste. JLB
2. The chemical senses: smell. JLB
3. The use of mouse genetics for gene function studies. AMZ
6. Auditory/vestibular systems. JLB
7. Somatic sensory system. JLB
8. Ciliopathies: Usher and Bardet Biedl. AMZ
9. Research seminar AMZ AMZ
10. Research seminar JLB JLB

Metodologia i organització general de l'assignatura

The course will be organized in 15 lectures, that will be accompanied by seminar sessions in which seminal or recent key papers in the field will be presented and discussed. Students will be expected to participate in the presentation and discussion of at least one research paper in these sessions, by working in teams. Research papers will be preselected by the instructors, and listed in the course syllabus ahead of time. Research articles, press notes or videos will be selected to complement and expand the lecture contents, matching the timing of the lecture and corresponding seminar, when possible.
Knowledge

Lecture contents and the object of seminar discussions.

Procedure

Final exam: answers to short questions covering the course syllabus (60%)
Participation in Seminars (40%)

Fonts d'informació bàsica

Textbooks:


FROM NEURON TO BRAIN (Nicholls, Martin, Wallace). Sinauer.


MEDICAL PHYSIOLOGY (Guyton and Hall). Elsevier.

THE FIRST STEPS IN SEEING. Sinauer.

Other materials:

Selected research papers from scientific journals.