

Workshop : Genetic Diseases of the Brain

Monday, 04 October 2010

Les résidents Accès sur inscription

EVENT_DESCRIPTION

Atelier IMéRA / INSERM dans le cadre de la résidence de [William Catterall](#) , professeur de pharmacologie à l'université de Washington, accueilli par l'IMéRA en partenariat avec l'INSERM.

IMéRA / INSERM Workshop, Marseille, Monday October 4 2010

Genetic Diseases of the Brain: Epilepsy, Ataxia, Migraine, and Bioethics

CNRS, salle de conférences Pierre Desnuelle, 31 ch. Joseph Aiguier (Entry free, no inscription required)

Nerve cells within the brain communicate with each other by electrical signals that are transmitted from cell to cell at synapses. Information entering the brain from sensory organs and commands leaving the brain to direct muscles and other organs are transmitted by electrical signals that move between nerve cells and other cell types at synapses. Electrical signaling is responsible for information processing and transmission in the brain. Regulation of the strength of synaptic communication, called 'synaptic plasticity', is also crucial for brain function. Short-term synaptic plasticity (milliseconds to seconds) encodes complex information for transmission from cell to cell. Long-term synaptic plasticity (minutes to days) is the first step in formation and retention of memories.

Electrical signaling and synaptic plasticity are disrupted in neurological and psychiatric diseases. The molecules that generate electrical signals and the molecular networks that control synaptic plasticity are an important focus of current research in neuroscience because of their essential role in information processing and transmission, learning, and memory in the brain and because of their impairment in disease. These essential brain processes are impaired in inherited forms of epilepsy, ataxia, migraine, and cognitive disturbances. Current research aims to understand the pathophysiology of these diseases and develop new approaches to their treatment, but these research areas raise important bioethical questions concerning gene-based, cell-based, and drug-based therapies. Genetic diseases of the nervous system present many dilemmas for diagnosis and treatment. These diseases are very rare, so it is uncertain whether the benefit of prenatal diagnosis outweighs the cost of genetic testing. It is also uncertain what level of severity of disease justifies genetic testing and clinical trials of novel therapies. Our program will begin with an introduction to this topic for a broad audience of scientists, physicians, other members of the university community, and interested members of the public. This will be followed by descriptions of cutting-edge research on three forms of genetic disease of the brain and an in-depth discussion of the bioethical issues that are raised by the emerging understanding of these genetic brain diseases.

Programme

14h00 - Pierre Livet (Université de Provence, IMéRA)

Research at the interface between biology and the humanities

14h10 - Michael Seagar (INSERM / Université de la Méditerranée)

Introduction to Electric Signaling and Synaptic Function in the Brain

15h00 - William A. Catterall (University of Washington, Seattle ; résident de l'IMéRA)

Insights into Pathophysiology and Therapy from a Mouse Model of Severe Myoclonic Epilepsy of Infancy

15h50 – 16h30 Coffee Break

16h30 Philippe Lory (CNRS / Université de Montpellier)

Ups and Downs of Calcium Channel Expression and Function in Epilepsy and Episodic Ataxia

17h10 Daniela Pietrobon (University of Padova)

Gain of Function of Cortical Excitatory Synaptic Transmission in Familial Hemiplegic Migraine

18h00 Hervé Chneiweiss (INSERM, Paris)

Bioethical Considerations in Diagnosis and Treatment of Inherited Brain Diseases

18h50 Overall Discussion

19h00 End of Workshop

CONTACT_PERSON

[Emmanuel Girard-Reydet](#)

EVENT_STARTINGPOINT

IMéRA, bâtiment "le Cube", site de l'ancien observatoire, plateau Longchamp, Marseille