EXPLORING THE CNS IN NEURODEVELOPMENTAL DISORDERS AND DISABILITIES

SECTION OF CHILD NEUROLOGY AND PSYCHIATRY
(Department of Clinical and Experimental Medicine University of Pisa)

and

IRCCS Fondazione Stella Maris For Developmental Clinical Neuroscience

Giovedì 1 Febbraio 2018 alle 17

Aula Magna, Scuola Medica
The research activity of the section of Child Neurology and Psychiatry of the University of Pisa is fully embedded with that of IRCCS Fondazione Stella Maris, where the Section is also physically located. Fondazione Stella Maris is a research hospital, belonging to a net of the Italian Ministry of Health. Major research interest is on the functional, neuroimaging and neurogenetic characteristics of the developmental age in sick conditions. Major infrastructures and technical expertise are in neuroimaging, functional analysis, molecular genetics, next-generation genomic and LC/MS/MS systems for protein peptide quantitation and biomarker verification, muscle morphology and validation of rare disease models in vitro and in vivo. In particular IMAGO7 Center, located at the IRCCS, offers unique opportunities for ultra-high field 7T-MR studies in adults and soon in children, and in animal models. Collaborative studies are in progress with other research laboratories in Pisa, in particular with IN and IFC of the CNR, around the topics described below. At the IRCCS, researchers are responsible for the development of new protocols for early diagnosis and treatment of neurodevelopmental disorders (NDD) in the fetus and the newborn, studying conditions affecting brain repair at those ages, and post-lesional cerebral reorganization secondary to motor, perceptual, and cognitive disorders, with the opportunity to investigate both model animals and young patients. Also, focus is on the genetic definition of NDD and neurodegenerative diseases of brain and muscle (NMD) occurring in childhood. Early diagnosis of rare neurogenetic conditions, definition of expression levels of sick genes in human cells or small vertebrates at the RNA and protein level, improved understanding of pathophysiology in order to identify disease modifiers and drug targets are major topics of the researchers at IRCCS.

In this seminar, after a general introduction around the above research facilities by Prof. Giovanni Cioni, few projects will be briefly presented by young researchers, in the field of neurogenetics and neuroimaging, as a glimpse on research activity in progress.

In the expanding field of neurodevelopmental and neurodegenerative disorders of childhood, modern neurogenetics offers new challenges in terms of more precise and immediate diagnosis and improved knowledge of the pathomechanisms. This might serve to prepare steps for future therapeutic aspects concerning neuroprotection or use of potential disease modifying treatments. We will discuss two new ways to investigate deeper the pathomechanisms of neurodegenerative disorders. Federica Morani, a Post-Doc in our lab, will recapitulate present efforts to study the pathophysiology of Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay (ARSACS), a debilitating neurodevelopmental and degenerative disorder of childhood characterized by retinal impairment and spastic-ataxia leading to difficulty in walking, speech impairment, and the use of a wheelchair. Dr. Morani will discuss the relationships between the potential chaperone function of sacsin (the sick protein in ARSACS) and its role in mitophagy, and how this research might improve our biological understanding on the ways sacsin operates in disease conditions.

Brain Magnetic Resonance Imaging (MRI) allows neuroscientists to study the living brain, providing detailed anatomical structural information, in healthy individuals or in subjects with disease. Several advanced MRI techniques, such a MR Spectroscopy, Diffusion MRI and other developed rapidly in the last decades, which have revolutionized neuroscience and have influenced neurological practice over years. 7T scanners permit a remarkable gain in terms of signal to noise ratio (SNR), which can be spent in terms of both spatial resolution (down to the order of hundreds of microns) and sensitivity, as well as in temporal resolution of dynamic phenomena. In this seminar Dr. Laura Biagi and Dr Simona Fiori will discuss possible technical benefits of the use of 7T-MRI and in particular for its application in functional studies (7T-fMRI) to the understanding of structure-function relationship, such as in plasticity mechanisms in congenital brain lesions Preliminary results will be presented on the use of 7T-fMRI in the study of somatosensory reorganization, by applying a task-dependent approach, by using a 7T-compatible pneumatic system developed ad hoc. Increase in spatial resolution, sensitivity and specificity at 7T allows a better localization of reorganized function in the native space of the subject, providing, for each single patient, specific information.