



Dottorato in Genetica, Biologia Molecolare e Cellulare
Università di Pavia - Dipartimento di Biologia e Biotecnologie "Lazzaro Spallanzani"

Course "Human Genetics and Pathology"

"Molecular mechanisms underlying human pathological conditions"

The course is held online

Organizers

Prof. Guglielmina N. Ranzani, Dipartimento di Biologia e Biotecnologie, Università di Pavia
Prof. Ornella Cazzalini, Prof. Monica Savio and Prof. Lucia Anna Stivala, Dipartimento di Medicina Molecolare, Università di Pavia
Dr. Elena Botta and Dr. Chiara Mondello Istituto di Genetica Molecolare L. L. Cavalli-Sforza, CNR, Pavia

Program

Prof. Barbara Bardoni, *Institute of Cellular and Molecular Pharmacology, CNRS UMR7275, Valbonne (FR)*

"Pathophysiology and therapeutic challenges of developmental brain disorders: the Fragile X syndrome as a paradigm"

The lectures will illustrate diagnostic criteria, comorbidities and causes of developmental brain disorders together with the animal models established for their study, the common interactomes and pathways underpinning their pathophysiology. Moreover, genetics, biology and therapeutic approaches for the Fragile X syndrome will be presented.

Monday 20 April 2020 - 14:30-16:30

"Developmental brain disorders"

Tuesday 21 April 2020 - 14:30-16.30

"The Fragile X syndrome"

Prof. Guglielmina N. Ranzani, *Dipartimento di Biologia e Biotecnologie, Università di Pavia*
Wednesday 22 April 2020 - 14:30-16:30

"The hereditary gastrointestinal cancer syndromes in the era of NGS"

Pathogenic germline variants in high-risk cancer genes have been implicated in 2-8% of all colorectal cancer and in 1-3% of all gastric cancer cases. The lecture will address how the advent of NGS and the use of broad gene panels not only have allowed the identification of new syndromes (i.e. new susceptibility genes and new carcinogenic mechanisms), but have also provided unexpected perspectives on well-known cancer syndromes and changed genetic-testing strategies for all of the familial gastrointestinal cancers.

Prof. Paola Vagnarelli, *College of Health and Life Science, Research Institute for Environment Health and Society, Brunel University, London (UK)*

“Genome organisation and diseases: cohesin and condensins”

Cohesin and condensins play an essential role in genome organisation. The lectures will address the molecular structure, biology and molecular mechanisms linking these two complexes to genome organisation in vertebrates. They will also present how defects in these complexes lead to a series of human diseases.

Thursday 23 April 2020 - 14:30-16:30

“Genome organisation: role of cohesin and cohesinopathies”

Friday 24 April 2020 - 14:30-16.30

“Genome organisation: role of condensins and condensinopathies”

Prof. Angelo Parini, *Institute of Metabolic and Cardiovascular Diseases, Toulouse (FR)*

“Cell senescence, aging and diseases: basic concepts and novel strategies for translational research.”

The lectures will deal with the general mechanisms of cell senescence. The recent findings on the molecular aspects of replicative (stromal cells) and post-mitotic (cardiomyocytes) senescence of cardiac cells will be addressed. We will also discuss the impact of cell senescence on the onset and progression of cardiac aging and failure. In the second part of the lectures, we will address the most relevant strategies to design appropriate animal cohorts to translate the results of basic research to humans, with a particular emphasis for the aging field.

Wednesday 29 April 2020 - 14:30-16:30

“From cell senescence to cardiac aging”

Thursday 30 April 2020 - 14:30-16:30

“Design of animal cohorts for translational research in the field of aging: inbred versus outbred mice”