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"

"Arturo Falaschi" room, Istituto di Genetica Molecolare – CNR, Pavia

Organizers
Prof. Guglielmina N. Ranzani, Dipartimento di Biologia e Biotecnologie, Università di Pavia
Prof. Ornella Cazzalini, Prof. Monica Savio and Prof. Lucianna Stivala, Dipartimento di Medicina Molecolare, Università di Pavia
Dr. Elena Botta and Dr. Chiara Mondello Istituto di Genetica Molecolare, 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 paly 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 cohesionpathies"
Friday 24 April 2020 – 14:30-16.30
"Genome organisation: role of condensins and condensinopathies"

Prof. Carlo Rivolta, *Institute of Molecular and Clinical Ophthalmology, Basel (CH)*
"Mendel 2.0; classical concepts of heredity re-evaluated at the scale of the full (human) genome"
The lectures will deal with fundamental characteristics of hereditary retinal degenerations, as a model for Mendelian diseases characterized by an elevated genetic heterogeneity. In addition, they will provide insights into the use of massively parallel sequencing technologies coupled with computer-assisted analyses of genomic data in current medical genetic research.
Monday 27 April 2020 - 14:30-16:30
"Molecular genetics of hereditary retinal degenerations"
Tuesday 28 April 2020 – 14:30-16.30
"Medical genetics in the digital age"

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