Dottorato in Genetica, Biologia Molecolare e Cellulare
Scuola di Dottorato in Scienze della Vita "Camillo Golgi" - Università degli Studi di Pavia

Course: **Molecular bases of hereditary and complex human diseases**

"Towards an understanding of the molecular basis of Mendelian and non-Mendelian" diseases

Organizers: C. Danesino, M. Paulli, L.A. Stivala, O. Zuffardi, P. Morbini, Dipartimento di Medicina Molecolare, Università di Pavia

Aula "A. Falaschi" IGM.CNR

February 9, 14.30-16.30
C. Danesino, University of Pavia
Lecture 1: Glycogen storage disease type II: from mutation to therapy (I)
Lecture 2: Glycogen storage disease type II: from mutation to therapy (II)

February 10, 16.30-18.30
C. Danesino, University of Pavia
Lecture 1: Phenotypic and genetic variability: the model of hereditary hemorragic telangiectasia
O. Zuffardi, University of Pavia
Lecture 2: Incomplete penetrance and variable expressivity: molecular basis

February 11, 14.30-17.30
F. Marini, University of Milan
Lecture 1: Genome instability syndromes: molecular mechanisms defective in Xeroderma pigmentosum and Fanconi anemia as key examples.
J. Maier, University of Milan
Lecture 2: TRMP7 and low magnesium: implication in endothelial dysfunction
L. Obici, Policlinico S. Matteo, Pavia
Lecture 3: Innate immunity and inflammation: insights from hereditary autoinflammatory diseases

February 12, 14.30-16.30
R. Ciccone, University of Pavia
Lecture 1: Copy number variation and point mutation in autism (I)
Lecture 2: Copy number variation and point mutation in autism (II)

February 13, 11:00-13:00
P. Morbini, University of Pavia
Lecture 1: Molecular bases of solid tumors: lung cancer
Lecture 2: Molecular bases of solid tumors: colon cancer

February 16, 14.30-16.30
S. Giglio, University of Pavia
Lecture 1: Genetic alterations in sporadic nephrotic syndrome and resistance to immunosuppression
A. Vanoli, Policlinico S. Matteo, Pavia
Lecture 2: Molecular bases of solid tumors: neuroendocrine tumors

February 17, 14.30-16.30
A. Brusco, University of Torino
Lecture 1: Spinocerebellar ataxia: phenotypic and genetic variability
M. Lucioni, Policlinico S. Matteo, Pavia
Lecture 2: Molecular bases of lymphoproliferative disorders

February 18, 14.30-16.30 *O. Zuffardi, University of Pavia*
O. Zuffardi, University of Pavia
Lecture 1: Somatic mosaicism and human diseases
G. Croci, University of Pavia
Lecture 2: Genes, infections, and lymphoproliferative disorders

February 19, 14.30-16.30
S. Giglio, University of Firenze
Lecture 1: Copy number variation in non-coding regions (I)
F. Novara, University of Pavia
Lecture 2: Copy number variation in non-coding regions (II)
A. Vetro, University of Pavia
Lecture 3: Copy number variation in non-coding regions (III)