ΙΤΕ/ΙΕΧΜΗ

ΣΕΜΙΝΑΡΙΟ

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ΟΜΙΛΗΤΗΣ: **Νικόλαος Μοσχονάς, Καθηγητής Γενικής Βιολογίας & Ιατρικής Μοριακής Γενετικής**
Εργαστήριο Γενικής Βιολογίας
Τμήμα Ιατρικής, Πανεπιστήμιο Πατρών

ΘΕΜΑ: **From the Human Genome Project (HGP) to Network Medicine**

ΤΟΠΟΣ: Αίθουσα Σεμιναρίων ΙΤΕ/ΙΕΧΜΗ

ΗΜΕΡΟΜΗΝΙΑ: **Τρίτη, 30 Ιουνίου 2015**

ΩΡΑ: **12:30**

ΠΕΡΙΛΗΨΗ

Nearly 25 years ago, the international enterprise for the analysis of the human genome, known as the Human Genome Project, started almost from scratch. Since then, rapidly evolving conventional as well as high-throughput technologies together with computational algorithms and tools for storage and in silico analysis of genetic information have revealed an enormous spectrum of impressive biological data, i.e. from the high resolution genetic and physical chromosome maps, to the complete human DNA sequence, and the determination of genetic variation characterizing thousands of human individuals and several population groups. Numerous positional cloning attempts and genome-wide association studies have currently, associated more than 6000 gene loci and gene variants to mendelian or multifactorial diseases permitting to a certain extent, valid prediction, early diagnosis and more effective treatment. Molecular signatures reflecting biological function at various levels of genetic reference have linked molecular and chromosomal aberrations to various types of genetic disease including cancer. However, due to the multiple levels of complexity characterizing the biological systems, there is still a huge gap for thorough understanding of the genotype/ phenotype functional relationships. Proteins, the fundamental contributors of cell processes, usually exert their biological role by interacting with other proteins ultimately forming a multi-protein network within the cell. In order to better understand biological function in health and disease, we work on the reconstruction and the analysis of a comprehensive human protein-protein network using a novel bottom-up knowledge-driven methodology for the integration of publicly available primary protein-protein interaction (PPI) data. PICKLE, our PPI knowledge base comprising of about 65% of the human

"reviewed" complete proteome and ~100.000 PPIs, has been used to reconstruct the human spliceosome protein interactome, and to project a highly reliable disease-causative gene/protein association dataset to the human PPI network. Analysis and further enrichment of the PPI-disease network may reveal important biological associations and molecular mechanisms underlying the molecular architecture of monogenic and multifactorial diseases.

N.K.Moschonas **(www.med.upatras.gr/en/Pages/people/dep.aspx?tID=21**) is Professor of Biology and Medical Molecular Genetics and the Director of the Department of General Biology, School of Medicine, U. of Patras. NKM graduated from the Department of Biology, U. of Patras (1975). As a Ph. D. candidate he was trained in molecular, developmental and evolutionary biology, and in protein chemistry at the Dept. of Biology, U. of Athens (1975) and the Dept. of Cellular & Developmental Biology, the Biological Laboratories, Harvard U. USA (1976-1979), under the supervision of Prof. F.C. Kafatos. As an EMBO postdoctoral fellow, he contributed with pioneer work on the molecular basis of β-thalassemia in early 80's, under the supervision of Prof. R. A. Flavell at the Division of Gene Structure & Expression, NIMR, MRC, Mill Hill, London, UK. From 1984 to 2006 he served as elected faculty member at the Department of Biology and Head of the Laboratory of Human Molecular Genetics, U. of Crete (Vice-Chairman of the Department, 2000-2004) with a joint appointment with the Institute of Molec. Biology and Biotechnology (IMBB)-FORTH.

NKM has participated in the coordination committees and management of a number of graduate programs (U. of Crete, U. Patras); he has organized and lectured in several undergraduate (U. of Crete, U. Patras) and graduate courses of Molec. Biology, Medical Molec. Genetics and Genomics, and Bioethics. (U. of Crete, U. of Patras, Athens U., U. of Thessaly, Aristotelian U. of Thessaloniki, and Demokrition U. of Thrace) He has supervised 13 Ph.D. and 18 MSc theses. Among several other academic and science management posts and activities, NKM has served as Deputy Member of the National Council for Research and Technology (2005-09) and as National Delegate in EU FP7 "Cooperation-Health" Program Committee (2006-10). NKM research interests are in the field of medical molecular genetics and genomics. His group participated in the international Human Genome Program from its very beginning and contributed to the genomics and genic map determination of human chromosome 10, and the analysis of disease-causative genes. His research aims to the elucidation of the molecular basis of genetic diseases by exploiting experimental and computational approaches including the use of in vitro and high-throughput technologies, cell and animal models and the analysis of big datasets to integrate the results of aberrant gene function as manifested at various levels of genetic reference. NKM's current interests include the development and analysis of a reliable integrated human protein and disease network, and its contribution for the determination of interrelated functional modules underlining the architecture of genetic diseases, with the aim to contribute in understanding disease pathophysiology, and suggest new effective therapeutics.