

Course Name: **Translational bigenomics – from the bedside to the bench and back again**

*IVth edition*

Date: **04-15 March 2013**

Organizer: Prof. Manuela Grazina

Email: [mgrazina@ci.uc.pt](mailto:mgrazina@ci.uc.pt), [mmgrazina@gmail.com](mailto:mmgrazina@gmail.com)

### Program and calendar:

#### **Monday – 04-03-2013**

**14:00** Introduction

**14:15** Genes in health and disease: ethics and legal issues

*Jorge Saraiva (MD, PhD, CHC, FMUC)*

**15:30** Basic concepts in Nuclear and Mitochondrial Genomics

*Manuela Grazina (PhD, FMUC)*

**17:30** Molecular Mechanisms of Gene Regulation

*Natércia Conceição (PhD, CCMAR-UALG)*

**18:30** Objectives and evaluation methods

**19:00** estimated ending time

#### **Tuesday – 05-03-2013**

**14:00** Genetic models of disease

*Leonor Cancela (PhD, CCMAR-UALG)*

**15:00** Update on methods for nucleic acids - Molecular Genetic analysis

*Manuela Grazina (PhD, FMUC)*

**16:30** Safety policy, certified rules, procedures and management of handling human samples

*Emília Telo (PhD, ACT)*

**17:30** Genetics in paediatric ophthalmology

*Eduardo Silva (MD, PhD, HUC, FMUC)*

**19:00** estimated ending time

#### **Wednesday – 06-03-2013**

**14:00** **Laboratory experiments designed for translational bigenomic approaches**

*Manuela Grazina (PhD, FMUC) assisted by Superior Technicians João Pratas and Maria João Santos (LBG, CNC-FMUC), and by Carolina Ribeiro (LBG, CNC/UC) and Daniela Luís (PhD student, FMUC)*

DNA and RNA Extraction/ Quantification

Planning and performing PCR, PCR-RFLP reactions

DNA electrophoresis

**19:00** estimated ending time

#### **Thursday – 07-03-2013**

**14:00** **Laboratory experiments designed for translational bigenomic approaches**

*Manuela Grazina (PhD, FMUC) assisted by Superior Technicians João Pratas and Maria João Santos (LBG, CNC-FMUC), and by Carolina Ribeiro (LBG, CNC/UC) and Daniela Luís (PhD student, FMUC)*

Planning and performing RT-PCR and real time-qPCR reactions

**19:00** estimated ending time

#### **Friday – 08-03-2013**

**Bioinformatics and Biostatistics as tools for translational genetics data**

**14:00** Genetic databases, primer design, tools for predicting pathogenicity *Manuela Grazina (PhD, FMUC)*

**15:00** Linkage analysis *Carolina Lemos (PhD, ICBAS, IBMC)*

**16:00** Association Studies *Carolina Lemos (PhD, ICBAS, IBMC)*

**17:30** **Special session:** Discussion and analysis of laboratory results

*Manuela Grazina (PhD, FMUC) and Lab collaborators*

**19:00** estimated ending time

**Monday – 11-03-2013**

**13:00** Diseases of bigenomic origin – Translation into Biochemistry

*Manuela Grazina (PhD, FMUC)*

**15:00** Inherited and sporadic neurodegenerative dementias

*Isabel Santana (MD, PhD, HUC, FMUC)*

**16:00** Impact of genetic predictive tests in Neurodegenerative diseases *Lina Ramos (MD, CHC)*

**17:00** Pre-implantation genetic diagnosis- Taylored babies: myth or reality? *Teresa Almeida Santos (MD, PhD, FMUC, CHUC)*

**18:00** **Special Session: OPEN DISCUSSION** Ethics and genetic testing, particularly predictive, in Diseases involving two genomes *(students are asked to do autonomic search and bring major guidelines based on peer-reviewed papers- one from each student)*

**19:30** estimated ending time

**Tuesday – 12-03-2013**

**14:00** Autism: a complex multigenic brain disorder

*Guiomar Oliveira (MD, PhD, FMUC, CHC)*

**15:00** Genetics in diagnosis and prognosis of pulmonary pathology

*Lina Carvalho (MD, PhD, FMUC, HUC)*

**16:30** Genetic compatibility and organ transplantation

*name to announce*

**18:00** estimated ending time

**Wednesday – 13-03-2013**

**14:00** A portrait of *Homo sapiens* as a young species: the mitochondrial DNA perspective

*Luisa Pereira (PhD, IPATIMUP)*

**15:00** Frontiers of the biology and genetics of aging. Telomers: the edge for pathology?

*Catarina Resende de Oliveira (MD, PhD, FMUC, CNC)*

**16:30** Thinking science in bigenomics area: brain storming *Manuela Grazina (PhD, FMUC)*

**19:30** estimated ending time

**Thursday – 14-03-2013**

**14:00** Mitochondrial genetic background predispose to disease and cancer

*Lee-Jun Wong (PhD, Baylor College of Medicine, Houston – Texas, USA)*

**15:00** Dual genome cross talk in health, disease, and cancer

*Lee-Jun Wong (PhD, Baylor College of Medicine, Houston – USA)*

**Special session:** Paper presentation and discussion I *(presentation by students; discussion with speakers)*

*Manuela Grazina (PhD, FMUC); Lee-Jun Wong (PhD, Baylor College of Medicine, Houston – Texas, USA)*

**16:30** Adult neurological manifestations of mitochondrial disease *Maria do Carmo Macário (MD, HUC)*

**17:30** Clinical manifestations of mitochondrial disease in children *Luísa Diogo (MD, PhD, FMUC, HPC-CHC)*

**19:30** estimated ending time

**Friday – 15-03-2013**

**14:00** Mitochondrial disorders holding nuclear genes responsible

*Lee-Jun Wong (PhD, Baylor College of Medicine, Houston – Texas, USA)*

**15:00** **Special session:** Paper presentation and discussion II *(presentation by students; discussion with speakers)*

*Manuela Grazina (PhD, FMUC); Lee-Jun Wong (PhD, BCM, Houston, USA)*

**16:30** **Seminar:** Next generation sequencing studies - **A giant step to the next generation molecular diagnosis of human genetic disorders**

*Lee-Jun Wong (PhD, BCM, Houston – Texas, USA)*

**17:30** **Special Session: MEET THE EXPERT** *(students are asked to delineate questions in groups, based on papers from the experts' team, and discuss with the experts)*

*Lee-Jun Wong PhD, (Baylor College of Medicine, Houston – Texas, USA); Manuela Grazina (PhD, FMUC)*

**18:30 – 19:00** Closing remarks