

## International Workshop on

# Genomic Disorders, Disease-Associated Chromosome Rearrangements and Position Effect

National Institute of Health “Dr. Ricardo Jorge”, Lisbon, Portugal

## Programme

### Friday 29<sup>th</sup> May 2009

8:00 Registration form in the Institute Foyer

9:00 Introduction and opening of the International Workshop (President of INSA.IP)

### 1st. Scientific session

Chair of the session:

9:15 - 10:00 **Characterising chromosomal rearrangement breakpoint associated with phenotypic abnormality**

David R FitzPatrick

Western General Hospital, Edinburgh, UK.

10:00 - 10:45 **Disease-associated rearrangements in neurological disorders**

Kerstin Kutsche

Universitätsklinikum Hamburg-Eppendorf, Hamburg, Germany.

10:45 - 11:15 Coffee break

11:15 - 12:00 **Bioinformatics of long-range gene regulation: enhancers, targets, development and disease**

Boris Lenhard

University of Bergen, Bergen, Norway

12:00 - 12:30 **Clinical implications of 13q31 deletion in two unrelated families. Biobanking and Biomolecular Resources Infrastructure (BBMRI)**

Béla Melegh

University of Pécs, Faculty of Medicine, Pécs, Hungary

György Fekete

Semmelweis Medical University Budapest, Hungary.

12:30 - 13:00 **Array CGH in clinical diagnosis**

Björn Menten

Ghent University Hospital, Ghent, Belgium

13:00 - 14:15 Lunch in the INSA

### 2nd. Scientific session

Chair of the session:

14:15 - 15:00 **Cis-regulatory control in disease and development: validation in humans and other models**

Veronica van Heyningen

Western General Hospital, Edinburgh, UK.

- 15:00 - 15:30 **Concerted involvement of synteny breaks, segmental duplications and increased transposon activity in malignancy-associated chromosome breakpoints**  
Stefan Imreh,  
Karolinska Institute, Stockholm, Sweden
- 15:30 - 16:00 **Array CGH in Clinical Research: Latest development of Technology and Applications**  
Yann Filaudeau  
Life Science Research and Applied Genomics - Agilent Technologies
- 16.00 - 16:15 Coffee break
- 16:15 -16:30 **UE Policies in the genomic area**  
Patrik Kolar  
Genomics and Systems Biology - European Commission
- 16:30 - 17:00 **Proteomic approaches to genomic disorders**  
Deborah Penque  
National Institute of Health Dr Ricardo Jorge, Lisbon, Portugal
- 17:00- 17:45 Poster Session
- 17:45- 18:15 Technical/scientific presentation
- 18:15 - 19:00 3 selected presentations from submitted abstracts (10 minutes each and 15 minutes for final discussion)
- 20.00 Conference diner (restaurant)

### **Saturday 30<sup>th</sup> May 2009**

#### **3rd. Scientific session**

Chair of the session:

- 9:00 - 09:45 **Beyond the open reading frame: Hidden mutations in human genetic disorders**  
Hans van Bokhoven  
Radboud University, Nijmegen Medical Centre, Nijmegen, The Netherlands
- 9:45 - 10:30 **4C technology: uncovering the multi-dimensional structure of the genome**  
Erik Splinter  
Erasmus Medical Center, Rotterdam, The Netherlands
- 10:30 - 11:00 Coffee break
- 11:00 - 11:30 **Genycell (Bluegenome platform)** –Technical/scientific presentation
- 11:30 - 11:50 A Portuguese invited speaker selected from submitted abstracts
- 11:50 - 12:00 Close of the workshop
- 12.00 **Close of conference**