

## Saturday, 17th November

*Chairs: Jorge Rocha and Luísa Mota-Vieira*

**09:00 Population Genetics of Common Diseases**

Anna di Rienzo, Department of Human Genetics,  
University of Chicago, USA

**09:40 Copy Number Variations in Health and Disease**

Xavier Estivill, Genes and Disease Program, Center for Genomic Regulation,  
Barcelona, Spain

**10:20 Coffee-Break**

*Chair: Alexandra Moreira*

**10:50 Orchestrating Gene Expression: mRNA Binding Proteins  
as Cellular Maestros**

Margarida Gama-Carvalho, Instituto de Medicina Molecular, Lisboa, Portugal

*Chairs: António Coutinho and Heloísa Santos*

**11:30 Teaching Genetics in Portugal**

Jorge Saraiva, Maria João Prata, Carolino Monteiro, Teresa Oliveira Marçal

**12:30 Lunch**

*Chair: Raquel Seruca*

**14:00 Comparative Genomics for the Study of Development, Evolution and  
Genetic Diseases**

José Luis Gómez-Skarmeta, Universidad Pablo de Olavide, Sevilla, Spain

**14:40 Closing Conference: Winner of the SPGH 2007 Award**

(to be announced)

**15:20 Award Ceremony**

**15:45 Adjourn**



SPGH Sociedade Portuguesa de Genética Humana

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## Reunião Anual SPGH

Porto 15 > 17 · Novembro · 2007

Centro de Cultura e Congressos  
da Ordem dos Médicos

### Apoios:



### Comissão organizadora:

Raquel Seruca  
Filipa Carvalho  
Céu Figueiredo

# Scientific Program

## Thursday, 15th November

- 09:00 **Secretariat Opening**
- 10:30 **Genetic Clubs** (Parallel Sessions)
- **Cytogenetics: “Cytogenetic Heteromorphisms”**  
Chairs: Madalena Ávila and Ana Cristina Duarte
- **Molecular Genetics: “Sequence Variants: Functionally Relevant?”**  
Chair: Carla Oliveira
- **Medical Genetics and Dysmorphology**  
Chairs: Margarida Reis Lima and Gabriela Soares
- 12:30 **Lunch**
- 14:30 **Opening Session**  
Raquel Seruca  
*Chairs: Filipa Carvalho and João Lavinha*
- 14:40 **Networks of Genes, Networks of Proteins, and Networks of Diseases**  
Han Brunner, Dept. Human Genetics, Nijmegen University Medical Center, The Netherlands
- 15:20 **Preimplantation Genetic Diagnosis - Designing Babies or a Useful Clinical Service**  
Alison Lashwood, Guy's Hospital, London, UK
- 16:00 **Coffee-Break**  
*Chair: Ana Fortuna and Isabel Marques Carreira*
- 16:30 **Genetics of Autism**  
Astrid Vicente, Instituto Nacional de Saúde Dr. Ricardo Jorge, Lisboa, Portugal
- 17:10 **Non Causal *de Novo* and Causal Inherited Chromosomal Imbalances: A Paradigm Shift in Cytogenetics**  
Joris Vermeesch, Center for Human Genetics, K.U.Leuven, Belgium
- 18:00 **General Assembly of the SPGH**

## Friday, 16th November | morning

- Chair: Sérgio Castedo*
- 09:00 **Spinal Muscular Atrophy: Therapeutic Perspectives and SMA Modifiers**  
Brunhilde Wirth, Institute of Human Genetics, University of Cologne, Germany
- 09:40 **Oral Communications I**
- OC1 An Efficient Protocol for Detection of Chromosomal Abnormalities in Spontaneous Abortions**  
S Doria, F Carvalho, C Ramalho, T Francisco, O Brandão, A Matias, M Sousa, A Barros
- OC2 Array Painting for Characterization of a Small Supernumerary Marker Chromosome**  
JB Melo, A Mascarenhas, M Pires, E Matoso, N Kosyakova, T Liehr, L Backx, J Vermeesch, IM Carreira
- OC3 Familial Hypercholesterolemia: Characterization of Families of Department of Medical Genetics, Santa Maria Hospital**  
O Moldovan, I Gomes, M Simão, S Martins, A Mourato, ML Antunes, LA Sousa, A Alves, M Bourbon, A Medeiros, H Santos, A Cabral, AM Gaspar, IM Gaspar
- OC4 Molecular Characterization of Familial Hypercholesterolaemia in Portuguese Patients**  
AM Medeiros, AC Alves, S Silva, M Bourbon
- 10:40 **Coffee-Break**  
*Chair: Peter Jordan*
- Oral Communications II**
- 11:00 **OC5 Alternative Splicing Controlled Expression of A Lymphocyte Cell Surface Receptor**  
M Castro, M Oliveira, R Nunes, S Fabre, R Barbosa, G Bismuth, A Moreira, A Carmo
- OC6 The Involvement of PABPC1 in Nonsense-Mediated Decay Inhibition of Human Beta-Globin Transcripts Carrying a Short Open Reading Frame**  
AL Silva, A Inácio, L Romão
- OC7 Study of Susceptibility to Breast Cancer Using Gene Expression Variation Analysis**  
AT Maia, AE Teschendorff, SF Chin, I Spiteri, L Jones, C Caldas, Bruce Ponder
- OC8 On the Track of the Out-of-Africa: Complete mtDNA Sequencing of L3 Haplotypes from Sudan**  
V Fernandes, JB Pereira, M Costa, C Afonso, F Alshamali, L Pereira
- 12:00 **Poster Visiting and Discussion**
- 12:45 **Lunch**

## Friday, 16th November | afternoon

- Chairs: Sérgio Dias and Helder Maiato*
- 14:00 **Hypomorphic Promoter Mutation in PIGM Causes Inherited Glycosyl-phosphatidylinositol Deficiency**  
António Almeida, Instituto Português Oncologia de Lisboa, Portugal
- 14:40 **Rac-1b and BRAF V600E Cooperate to Maintain Transformation of Colorectal Cancer Cells**  
Paulo Matos, Instituto Nacional de Saúde Dr. Ricardo Jorge, Lisboa, Portugal
- 15:10 **Epigenetics for Biotechnology, Developmental Control and Mood Regulation**  
Didier Trono, School of Life Sciences, Global Health Institute, EPFL, Lausanne, Switzerland
- 15:50 **Coffee-Break**  
*Chairs: Patrícia Maciel and António Jacinto*
- 16:20 **Nerve Regeneration: From Neuropathies to Leukodystrophies**  
Mónica Sousa, Instituto de Biologia Molecular e Celular, Porto, Portugal
- 17:00 **The Role of Hox Genes in Skeletal Development**  
Moisés Mallo, Instituto Gulbenkian de Ciência, Oeiras, Portugal
- 17:40 **Oral Communications III**
- OC9 P-cadherin Induces Breast Cancer Cells Motility and Stimulates Cell Invasion Through Secretion of Pro-Invasive Factors**  
AS Ribeiro, J Paredes, AL Correia, R Seruca, F Schmitt
- OC10 Loss of Expression of E-Cadherin Juxtamembrane Missense Mutations: a Task for the Proteasome?**  
J Simões-Correia, R Seruca, J Van Helgel, F van Roy, G Suriano
- OC11 Novel Mutations of SPG4 in Portuguese Patients with Spastic Paraplegia**  
AM Lopes, C Matos, L Fleming, JM Loureiro, I Alonso, I Silveira, P Coutinho, J Sequeiros
- OC12 The First Portuguese Report on Clinical Characterization and Mutation Analysis of the NIPBL Gene in 30 Patients with Cornelia de Lange Syndrome**  
C Dias, J Oliveira, E Costa, J Silva, M Martins, AM Fortuna, I Soares-Silva, AB Sousa, T Kay, H Santos, A Medeira, I Cordeiro, T Lourenço, L Nunes, R Santos, MR Lima
- 20:30 **Conference Dinner**