

Scientific Program

Time	Thursday, 10 th November
10:00 - 11:00	Secretariat opening and setting of commercial exhibition
11:00 - 13:00	Thematic Clubs (Parallel Sessions) <p>Cytogenetics: Mosaicism Chair: Sofia Dória</p> <p>Molecular Genetics: Consequences of the recent law concerning personal genetic information and health information to the molecular genetics laboratory Chair: Luísa Mota Vieira</p> <p>Clinical Genetics and Dysmorphology: Recent advances in two old situations - Cornelia de Lange Syndrome and CHARGE Syndrome Chairs: J Carlos Ferreira/Ana Medeira</p>
13:00 - 14:00	Lunch
14:00 - 15:00	Registration and poster exhibition set up
	Opening Session
15:00 - 15:20	Introductory remarks (SPGH, ProCura and Health Ministry)
15:20 - 16:00	Plenary Lecture Jason H Moore (Dartmouth-Hitchcock Medical School, Lebanon, NH): Systems Biology in Biomedicine and Health Care – A global view of epistasis
16:00 - 16:30	Coffee and Poster Viewing
PARALLEL SESSION - SPGH (Room A)	
16:30 - 17:30	Oral Communications I <p>OC1 Clinical hypochondroplasia in a family caused by a heterozygous double mutation in FGFR3 encoding gly380lys Heloísa G. Santos, Rosário Almeida, Helena C. Fernandes, Andrew O.M. Wilkie</p> <p>OC2 A study of 87 extended HLA haplotypes in HFE-C282Y homozygous hemochromatosis subjects: relationship to the genetic control of CD8+ T-lymphocyte numbers and severity of iron overload Eugénia Cruz, Jorge Vieira, Susana Almeida, Rosa Lacerda, Andrea Gardner, Helena Alves, Graça Porto</p> <p>OC3 Altered BMP signalling in a cellular model of oculopharyngeal muscular dystrophy Patrícia Calado, Sandra Caldeira, Maria do Carmo Fonseca</p> <p>OC4 Gene expression profiling in vLINCL CLN6-deficient fibroblasts: insights into pathobiology Teixeira CAF, Lin S, Mangas M, Quinta R, Bessa CJP, Ferreira C, Sá Miranda MC, Boustany R-M N., Ribeiro MG</p>
17:30 - 18:30	Round table discussion on R&D in Human Genetics: Expectations, constrains and priorities Chair: João Lavinha Francisco Corte-Real (INML), Rui Guimarães (COTEC), Paulo Fidalgo (IPO) and Lourdes Fonseca (APH)
PARALLEL SESSION - ProCura (Room B)	
	Advances in Proteomics I: technologies and methodologies Chair: Ana V Coelho
16:30 - 17:00	Jean Andersen (Univ Odense): SILAC labelling to investigate the function and dynamics of large multi-protein complex

17:00 - 17:30 Stephen Tate (Stephen Tate, Applera Europe) **MALDI Imaging: A New Technique for Drug Distribution Studies**

17:30 - 18:00 Holger Husi (Univ of Edinburgh): **PADB (proteomic analysis dabase), a novel proteomic resource for large scale systems biology analysis**

18:00 - 18:30 **Oral Communications II**

OC5 Protein characterization by mass spectrometry

Ana V Coelho

OC6 Identification and characterization of soil bacterium using mass spectrometry approach: fingerprinting *Sinorhizobium* cell surface proteins by MALDI-TOF mass spectrometry

Ferraz Franco C., Alves P. I., Faroleira P., Crespo M.T. Barreto, Coelho A. V.

19:00

Welcome Cocktail

Time	Friday, 11th November
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9:00 - 9:45 **Plenary Lectures**

Thomas Conrads (National Cancer Institute at Frederick, MD): **Clinical proteomics: Identification and characterization of the antiproliferative factor from the urine of interstitial cystitis patients**

Chair: Deborah Penque

9:45 - 10:30 Rui Reis (Univ Minho): **Investigação em células estaminais e terapia celular: factos e ficções**

Chair: Ana Fortuna

10:30 - 11:15

Coffee and Poster Viewing

PARALLEL SESSION - SPGH (Room A)

Emerging Technology in Human Genetics

Chair: Isabel Carreira

11:15 - 12:00 Niels Tommerup (Copenhagen Univ): **Array CGH in microdeletion / duplication detection**

12:00 -13:00 **Oral Communications III**

OC7 Hibridização genómica comparativa: contribuição para o estudo de produtos de abortamento

Sofia Dória, Filipa Carvalho, Alexandra Matias, Manuel R Teixeira, Franklin Ribeiro, Lurdes Torres, Mário Sousa, Alberto Barros

OC8 Evaluation of numerical chromosome aberrations in glioblastoma tumours as revealed by interphase fluorescence in situ hybridization

Ana Luísa Vital, Maria Dolores Taberner, Olinda Rebelo, Hermínio Tão, Raimundo Fernandes, Maria Celeste Lopes, Alberto Orfão

OC 9 Diagnóstico pré-natal de aneuploidias cromossómicas utilizando um novo método molecular quantitativo

Filipa Carvalho, Alexandra Matias, Josefa Jaume Barceló, Sofia Dória, Mário Sousa, Sérgio Castedo, Alberto Barros

OC10 Effective isolation, purification and cryopreservation of human adult testicular progenitor germ cells

Sá R, Sousa M, Cremades N, Escoriza JC, Silva J, Fernandes S, Carvalho F, Barros A

PARALLEL SESSION - ProCura (Room B)

Proteomics: uncovering biological mechanisms I

Chair: Candido Pinto Ricardo

11:15 - 11:45 Jesús V. Jorrín Novo (Univ Córdoba, Spain): **Plant proteomics: Technical platforms and current research projects**

11:45 - 12:15 C Pinto Ricardo (ITQB, Oeiras): **Proteomic Studies at the Plant Biochemistry Laboratory (ITQB)**

12:15 - 12:45 **Oral Communications IV**

OC11 Proteomics of the plant response to boron deficiency

Alves M, Francisco R, Martins I, Ricardo C P

OC12 Poplar, chestnut and grapevine linked through -omics

Sílvia Ferreira, Andreia Figueiredo, Fernando Pessoa, Dora Batista, Mónica Sebastiana, Bartolomeu Acioli, Maria Salomé Pais

13:00 - 14:00

Lunch Break

PARALLEL SESSION - SPGH (Room A)

Neurogenetics and behaviour genetics

Chair: M Luís Cardoso

14:00 - 14:45 Mário Gomes-Pereira (Hôpital Necker Enfants Malades, Paris): **DNA trinucleotide repeats and human disease: what goes up, must come down**

14:45 - 15:40 Oral Communications V

OC13 Evidence for epistasis in the determination of hyperserotonemia and in autism etiology

A.M. Coutinho, G. Oliveira, I. Sousa, T. Morgadinho, C. Fesel, T.R. Macedo, C. Bento, M. Martins, C. Marques, A. Ataíde, T.S. Miguel, L. Borges, A.M. Vicente

OC14 33 famílias com Neurofibromatose tipo 1 (NF1): a casuística da consulta do Instituto de Genética Médica

Jorge Pinto-Basto, Gabriela Soares, Cristina Dias, Miguel Rocha, Márcia Martins, Ana Maria Fortuna, Margarida Reis Lima

OC15 Inactivation status of *PCDH11X*: sexual dimorphism in gene expression levels in brain

Alexandra Lopes, Norman Ross, James Close, Adam Dagnall, Timothy J. Crow, António Amorim

OC16 Associação em adolescentes obesas do polimorfismo genético 5-HTTVNTR do gene do transportador da serotonina, das concentrações de serotonina e leptina e da actividade da MAO B

Pilar Levy, Joana Ferreira, Sara Neves, Helena Fonseca, João Gomes-Pedro, Manuel Bicho

PARALLEL SESSION - ProCura (Room B)

Proteomics: therapeutic target & biomarker discovery

Chair: Edgar da Cruz e Silva

14:00 - 14:30 Györg Marko-Varga (Univ Lund): **Discovery of Peptides and Proteins within Disease: Surveillance Strategies**

14:30 - 15:00 Anthony Gonçalves (Institut de Cancérologie de Marseille): **SELDI-TOF MS-based proteomics of human breast cancer: from prognostic serum biomarkers to a protein portrait of tumours**

15:00 - 15:30 Edgar da Cruz e Silva (Univ Aveiro): **Proteomics and yeast two-hybrid characterization of protein phosphatase interactomes and identification of potential targets for signal transduction therapeutics**

15:30 - 15:40 Oral Communications VI

OC17 Mass spectrometry identification on proteins from mouse proximal bronchial epithelial cells obtained by laser-microdissection microscopy

I. Carvalho-Oliveira, T. Luider, J. Demmers, D. Penque, B.J. Scholte

15:40 - 16:00

Coffee and Poster Viewing

PARALLEL SESSION - SPGH (Room A)

Population genetics of men, domesticates and its parasites

Chair: António Amorim

16:00-16:45 Rosalind Harding (Oxford Univ): **Beyond phylogeny: evolutionary analysis of a mosaic pathogen**

16:45-18:00 Oral Communications VII

OC18 Mitochondrial DNA lineages in five Portuguese dog breeds and a mixed-breed population: does diversity reveal different artificial selection pressures?

Barbara van Asch, Filipe Pereira, António Amorim

- OC19 Estudo da variabilidade genética do Arquipélago da Madeira recorrendo ao cromossoma Y e ao DNA mitocondrial**
Ana T. Fernandes, Rita Gonçalves, Alexandra Rosa, António Brehm
- OC20 Caracterização da diversidade genética da população Açoriana através da análise de 13 microssatélites**
Branco CC, Pacheco PR, Cabral R, de Fez L, Peixoto BR, Mota-Vieira L
- OC21 Genetic modifiers of morbidity in sickle cell disease**
Rute Martins, Anabela Morais, Alexandra Dias, Lúgia Braga, Isabel Soares, Cristiana Rolão, Baltazar Nunes, Isabel Picanço, Susana Gomes, Armandina Miranda, Teresa Seixas, Fernanda Rego, José L. Ducla-Soares, Gabriel Olim, Luísa Romão, Paula Faustino
- OC22 Machado-Joseph disease is present worldwide and may have an ancestral Japanese origin: haplotype backgrounds, age estimations and routes explaining current frequencies**
S Martins, C Gaspar, F Calafell, I Silveira, V Wong, M Hsieh, B-W Soong, L Tranebjaerg, E Brunt, GA Nicholson, S Tsuji, L Jardim, M Watanabe, O Riess, N Wood, H Sasaki, L Ranum, A Brice, G Rouleau, P Coutinho, A Amorim, J Sequeiros

18:00-19:00 SPGH Membership Meeting

PARALLEL SESSION - ProCura (Room B)

Proteomics: uncovering biological mechanisms II

Chair: Celso Cunha

- 16:00 - 16:30** Concha Gil (Univ Complutense, Madrid): **Proteomics and Bioinformatics present Novel Diagnostic, Prognostic and Therapeutic Candidates for Systemic Candidiasis.**
- 16:30 - 17:00** J Pablo Albar (CNB-CSIC, Madrid): **A Computational Framework for label-free MS-based proteomic biomarker discovery.**
- 17:00 - 18:00 Oral Communications VIII**
- OC23 Proteome differences caused by the transient expression of the hepatitis delta antigens in a Human Liver cell line**
S. Mota, D. Penque, M. Mendes, A. Coelho, C. Cunha
- OC24 The role of melanization on immune response to Plasmodium in mosquito hemolymph**
S. Ramos, A. Gabriel, V.E. do Rosário, H. Silveira
- OC25 Influence of the HDV genomic and antigenomic RNA expression in human liver cells proteome**
M. Mendes, S. Mota, D. Penque, A. Coelho, C. Cunha
- OC26 Salivary protein composition: How it influences *in vitro* pellicle composition and its susceptibility for dental caries**
Rui Vitorino, Maria J. C. Lobo, José A. R. Duarte, A.J. Ferrer-Correia, Pedro Domingues, Francisco Amado

20:00

Conference Dinner

Time

Saturday, 12th November

- 9:00 - 9:45 Comparative genomics**
Daniel G Bradley (Trinity College Dublin): **The genetics of domestication, a comparative approach**
Chair: Luisa Azevedo
- 9:45 - 10:30 Oral Communications IX**
- OC27 Somatic and intergenerational instability of expanded CAG repeat in a transgenic mouse model of Machado-Joseph disease**
Anabela Silva-Fernandes, Maria do Carmo Costa, Tiago Ferreira, Patrícia Maciel
- OC28 Expression profile of splicing factors during myogenesis**
Anita Gomes, Sandra Caldeira, Nuno Morais, Maria Carmo-Fonseca
- OC29 Mice as models for phylogenetic studies**
Ana Goios, Luísa Pereira, Molly Bogue, Vincent Macaulay, António Amorim

10:30 - 11:00

Coffee and Poster Viewing

11:00 - 12:00 **Cancer genetics**

Oral Communications X

Chair: Guida Boavida

OC30 Regulation of alternative splicing of the small GTPase Rac1

Vânia Gonçalves, Peter Jordan

OC31 Identificação e mapeamento da interacção entre o TCF4 e o Grg5

Ângela M. S. Costa, Luís Teixeira da Costa

OC32 A deficiência em poli(ADP-ribose)polimerase-1 in vivo causa um aumento de deleções/inserções em resposta a lesões alquilantes do DNA

Henriqueta Louro, Maria João Silva, Inês Pinheiro, Paula Costa, Ana Carla Sousa, Anabela Dias, Maria Guida Boavida

12:00 - 12:30 **Closing conference: Winner of the SPGH 2005 Award** (to be announced)

Chair: J Carlos Ferreira

12:30 - 13:00 **Award Ceremony and Closing Session** (SPGH and Science, Technology and Higher Education Ministry)
