



Sociedade Portuguesa de Genética Humana

21st Annual Meeting | 16 – 18 November 2017

Venue | *Hotel ALDEIA DOS CAPUCHOS, GOLF & SPA, Largo Aldeia dos Capuchos, Caparica, Portugal*

Scientific Program

Thursday, November 16

- 9:30** **Registration Opening**
- 10:00** **Cytogenetics and Molecular Genetics Club**
Hildeberto Correia
Sílvia Serafim
João Gonçalves
Patrícia Theisen
Luís Vieira
Catarina Silva
 [Instituto Nacional de Saúde Doutor Ricardo Jorge (INSA), Lisboa, Portugal]
- Medical Genetics and Clinical Dysmorphology Club**
Patrícia Dias
Vera Silva Lopes
Catarina Machado
André Travessa
Mariana Sá
 [Serviço de Genética Médica, Centro Hospitalar Lisboa Norte (CHLN), Hospital de Santa Maria (HSM), Lisboa, Portugal]
- 14:00** **Opening and Welcome**
 Direção da SPGH
 Conselho Diretivo do INSA
- 14:15** **Personalized Medicine for Cystic Fibrosis**
Margarida Amaral, Faculdade de Ciências, Universidade de Lisboa, Lisboa, Portugal
Chair: João Lavinha
- 15:00** **DYNAMICS OF GENOME ARCHITECTURE**
Chairs: Joana Barbosa de Melo; Sérgio Bernardo Sousa
- Interpretation of Structural Variations in a Regulatory Context**
Stefan Mundlos, Max Planck Institute for Molecular Genetics, Berlin, Germany
- Spectrum of Structural Genomic Abnormalities in Subjects Carrying Putative Disease-Associated Chromosome Rearrangements and their Pathogenic Implications**
Dezso David, INSA, Lisboa, Portugal
- Chromatin Reorganization Events in Endothelial Cells Associated with Pulmonary Arterial Hypertension**
Armando Reyes Palomares, European Molecular Biology Laboratory (EMBL), Heidelberg, Germany
- 16:30** **Coffee Break / Poster Viewing**

17:00 Selected Oral Communications I (1-6)

Chairs: Susana Fernandes; Sofia Dória

18:00 Genetic Counselling and Personalized Medicine in Rare Disorders

Corporate Panel Discussion (Shire)

Moderator: Maria do Céu Machado, Presidente do Infarmed, Lisboa, Portugal

Fabry's disease: phenotypic heterogeneity and therapeutic implications

Patrício Aguiar, Centro de Referência em Doenças Metabólicas, CHLH-HSM, Faculdade de Medicina da Universidade de Lisboa, Lisboa, Portugal

Fabry's disease: issues in genetic counseling

Oana Moldovan, Serviço de Genética Médica, CHLN-HSM, Lisboa, Portugal

Genetic Counselling: the psychological dimensions in the individual and family. A reflection from the clinic.

Alexandra Leonardo, Serviço de Genética Médica, CHLN-HSM, Lisboa, Portugal

18:45 SPGH Assembly



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Friday, November 17

- 8:45** **Selected Oral Communications II (7-12)**
Chairs: Carla Oliveira; João Fernando Silva
- 9:45** **Nusinersen (Spinraza™): The First FDA/EMA-Approved Treatment for SMA**
Adrian Krainer, Cold Spring Harbor Laboratory, Cold Spring Harbor, NY, USA
Chair: Luísa Romão
- 10:30** **Coffee Break / Poster Viewing**
- 11:00** **CANCER GENETICS**
Chairs: Paulo Matos; Peter Jordan
- 11:00** **Networks of Alternative Splicing Regulation in Cancer**
Juan Valcarcel, Centre for Genomic Regulation, The Barcelona Institute of Science and Technology, Barcelona, Spain
- 11:30** **RNA Binding Proteins in Cancer Progression**
Fatima Gebauer, Centre for Genomic Regulation, The Barcelona Institute of Science and Technology, Barcelona, Spain
- 12:00** **Selected Oral Communications III (13-18)**
Chairs: Rosário Pinto Leite; Ana Medeira
- 13:00** **Lunch**
- 14:15** **TOWARDS PERSONALIZED MEDICINE IN ONCOLOGY**
Chairs: Manuel Teixeira; Juliette Dupont
- Classification of Mutations in the Exonuclease Domain of POLE**
Claire Palles, Wellcome Trust Centre for Human Genetics, University of Oxford, Oxford, UK
- Targeting Cancers using Individual Systems Medicine**
Kristen Wennerberg, Institute for Molecular Medicine Finland, Nordic EMBL Partnership for Molecular Medicine, University of Helsinki, Helsinki, Finland

15:15 MECHANISM-GUIDED THERAPIES FOR GENETIC DISORDERS

Chairs: Ana Sousa; Glória Isidro

Translational Medicine in Familial Hypercholesterolemia – from Phenotype to Genotype to Treatment

Mafalda Bourbon, INSA, Lisboa, Portugal

Development of RNA Based Approaches to Exploit Alternative Therapies for Lysosomal Storage Diseases

Sandra Alves, INSA, Porto, Portugal

16:15 Coffee Break / Poster Viewing**16:45 Genetic Counselling and Personalized Medicine in Cancer**

Corporate Panel Discussion (Astrazeneca)

Moderator: Ana Berta Sousa, Serviço de Genética Médica, CHLN-HSM, Lisboa, Portugal

Mainstreaming Cancer Genetics

Angela George, Cancer Genetics Unit, The Royal Marsden, London, UK

Commentary: The Portuguese Situation and Where to Go from Here

Manuel Teixeira, Serviço de Genética, Instituto Português de Oncologia do Porto Francisco Gentil, Porto, Portugal

17:30 Panel Discussion “Challenges in Personalized Medicine”

Moderator: Astrid Vicente, INSA, Lisboa, Portugal

Further Knowledge towards Improved Precision

João Lavinha, INSA, Lisboa, Portugal

Genetic Tests and Personalized Medicine in Hospital Settings

Luisa Mota Vieira, Hospital do Divino Espírito Santo, Ponta Delgada, Portugal

ICT and Data-driven Medicine: Challenges and Solutions

Tiago Guerreiro, Faculdade de Ciências, Universidade de Lisboa, Lisboa, Portugal

Personalized Medicine Today: Who Knows What?

Luciana Costa, INSA, Lisboa, Portugal

Personalized Medicine in the R & I Agenda for Health, Clinical and Translational Research

Marta Abrantes, Fundação para a Ciência e a Tecnologia, Lisboa, Portugal

19:00 City Tour**21:00 Conference Dinner**



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Saturday, November 18

- 9:00** **Selected Clinical Cases for Oral Communication (1-10)**
Chairs: Lina Ramos; Isabel Cordeiro
- 10:00** **Bioethics Debate**
Medicina Personalizada na Era Genómica
Personalized Medicine in the Era of Genomics
Moderador: [Heloísa Santos](#), Presidente Comissão Bioética SPGH, Lisboa, Portugal
- Considerações Éticas e Científicas da Medicina Personalizada**
Scientific and Ethical Issues in Personalized Medicine
[Célia Ventura](#), INSA, Lisboa, Portugal
- A medicina Personalizada Face à Legislação Portuguesa**
Portuguese Legal Issues in Personalized Medicine
[André Pereira](#), Faculdade de Direito da Universidade de Coimbra, Coimbra, Portugal
- 11:00** **Coffee Break / Poster Viewing**
- 11:30** **Genome Editing with Programmable Nucleases**
[Dana Carroll](#), University of Utah School of Medicine, Salt Lake City, UT, USA
Chair: Rosário Santos
- 12:15** **Basic and Clinical Research Awards Ceremony**
- 12:25** **SPGH Award Conference**
- 12:55** **Closing Session**
Conselho Diretivo do INSA
Direção da SPGH