



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 Patrícia Theisen Catarina Silva [Instituto Nacional de Saúde Doutor Ricardo Jorge (INSA), Lisboa, Portugal]	Medical Genetics and Clinical Dysmorphology Club Patrícia Dias 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 <i>Chair: João Lavinha</i>	
15:00	DYNAMICS OF GENOME ARCHITECTURE <i>Chairs: Joana Barbosa de Melo; Sérgio Bernardo Sousa</i>	
15:00	Interpretation of Structural Variations in a Regulatory Context Stefan Mundlos , Max Planck Institute for Molecular Genetics, Berlin, Germany	
15:30	Spectrum of Structural Genomic Abnormalities in Subjects Carrying Putative Disease-Associated Chromosome Rearrangements and their Pathogenic Implications Dezso David , Instituto Nacional de Saúde Doutor Ricardo Jorge, Lisboa, Portugal	
16:00	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 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
- 14:15** **Classification of Mutations in the Exonuclease Domain of POLE**
Claire Palles, Wellcome Trust Centre for Human Genetics, University of Oxford, Oxford, UK
- 14:45** **Targeting Cancers using Individual Systems Medicine**
Krister 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
- 15:15** **Translational Medicine in Familial Hypercholesterolemia – from Phenotype to Genotype to Treatment**
Mafalda Bourbon, Instituto Nacional de Saúde Doutor Ricardo Jorge, Lisboa, Portugal
- 15:45** **Development of RNA Based Approaches to Exploit Alternative Therapies for Lysosomal Storage Diseases**
Sandra Alves, Instituto Nacional de Saúde Doutor Ricardo Jorge, 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
(to be completed)
- 17:30** Panel Discussion “**Personalized Medicine**”
Moderator: Astrid Vicente, Instituto Nacional de Saúde Doutor Ricardo Jorge, Lisboa, Portugal
(to be completed)
- 18:15** **Corporate Symposium** (Soquímica)
- 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](#), Instituto Nacional de Saúde Doutor Ricardo Jorge, 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