



## 21<sup>st</sup> Annual Meeting | 16 – 18 November 2017

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

### Scientific Programme

#### Thursday, November 16

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|--------------|--|---|
| <b>9:30</b>  | <b>Registration Opening</b>  |   |
| <b>10:00</b> | <b>Cytogenetics and Molecular Genetics Club</b>  | <b>Medical Genetics and Clinical Dysmorphology Club</b> |
| <b>14:00</b> | <b>Opening and Welcome</b><br>Luísa Romão, SPGH President<br>Ana Sousa, SPGH Secretary<br>Rosário Pinto Leite, SPGH Treasurer  |   |
| <b>14:15</b> | <b>Personalized Medicine for Cystic Fibrosis</b><br><a href="#">Margarida Amaral</a> , Faculdade de Ciências, Universidade de Lisboa   |   |
| <b>15:00</b> | <b>Interpretation of Structural Variations in a Regulatory Context</b><br><a href="#">Stefan Mundlos</a> , Max Planck Institute for Molecular Genetics, Berlin, Germany  |   |
| <b>15:30</b> | <b>Spectrum of Structural Genomic Abnormalities in Subjects Carrying Putative Disease-Associated Chromosome Rearrangements and their Pathogenic Implications</b><br><a href="#">Dezso David</a> , Instituto Nacional de Saúde Doutor Ricardo Jorge, Lisboa |   |
| <b>16:00</b> | <b>Coffee Break / Poster Viewing</b>   |   |
| <b>16:30</b> | <b>Chromatin Reorganization Events in Endothelial Cells Associated with Pulmonary Arterial Hypertension</b><br><a href="#">Armando Reyes Palomares</a> , EMBL, Heidelberg, Germany   |   |
| <b>17:00</b> | <b>Selected Oral Communications I (6) (8+2 min communication)</b>  |   |
| <b>18:00</b> | <b>Genetic Counselling and Personalized Medicine in Rare Disorders</b><br>Corporate Panel Discussion (Shire)<br><i>(to be confirmed)</i>   |   |
| <b>18:45</b> | <b>SPGH Assembly</b>   |   |



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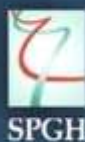
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### Scientific Programme

#### Friday, November 17

- 9:00** Selected Oral Communications II (9)
- 10:30** **Nusinersen (Spinraza™): The First FDA/EMA-Approved Treatment for SMA**  
**Adrian Krainer**, Cold Spring Harbor Laboratory, NY, USA
- 11:15** **Coffee Break / Poster Viewing**
- 11:45** **Networks of Alternative Splicing Regulation in Cancer**  
**Juan Valcarcel**, Centre for Genomic Regulation, The Barcelona Institute of Science and Technology, Barcelona, Spain
- 12:15** **RNA Binding Proteins in Cancer Progression**  
**Fatima Gebauer**, Centre for Genomic Regulation, The Barcelona Institute of Science and Technology, Barcelona, Spain
- 12:45** **Lunch**
- 14:00** **Classification of Mutations in the Exonuclease Domain of POLE**  
**Claire Palles**, Wellcome Trust Centre for Human Genetics, University of Oxford, Oxford, UK
- 14:30** **Targeting Cancers using Individual Systems Medicine**  
**Krister Wennerberg**, Institute for Molecular Medicine Finland, Nordic EMBL Partnership for Molecular Medicine, Finland
- 15:00** **Translational Medicine in Familial Hypercholesterolaemia – from Phenotype to Genotype to Treatment**  
**Mafalda Bourbon**, Instituto Nacional de Saúde Doutor Ricardo Jorge, Lisboa
- 15:30** **Development of RNA Based Approaches to Exploit Alternative Therapies for Lysosomal Storage Diseases**  
**Sandra Alves**, Instituto Nacional de Saúde Doutor Ricardo Jorge, Porto
- 16:00** **Coffee Break / Poster Viewing**
- 16:30** Panel Discussion “**Personalized Medicine**”  
**Moderator:** Astrid Vicente, Instituto Nacional de Saúde Doutor Ricardo Jorge, Lisboa

- 17:20**      **Genetic Counselling and Personalized Medicine in Cancer**  
Corporate Panel Discussion (Astrazeneca)  
**Moderator:** Ana Berta Sousa, Hospital de Santa Maria/CHLN, Lisboa
- 18:00**      Corporate Symposium (Soquímica)  
*(to be confirmed)*
- 20:30**      **Conference Dinner**



# Sociedade Portuguesa de Genética Humana

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## Scientific Programme

**Saturday, November 18**

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|--------------|---|
| <b>9:00</b>  | <b>Selected Oral Communications III (6)</b>   |
| <b>10:00</b> | <p><b>Bioethics Debate</b><br/> <b>Medicina Personalizada na Era Genómica</b><br/> <b>Personalized Medicine in the Era of Genomics</b><br/> <b>Moderador: <a href="#">Heloisa Santos</a></b>, Presidente Comissão Bioética SPGH</p> <p><b>Considerações Éticas e Científicas da Medicina Personalizada</b><br/> <b>Scientific and Ethical Issues in Personalized Medicine</b><br/> <b><a href="#">Célia Ventura</a></b>, Instituto Nacional de Saúde Doutor Ricardo Jorge, Lisboa</p> <p><b>A medicina Personalizada Face à Legislação Portuguesa</b><br/> <b>Portuguese Legal Issues in Personalized Medicine</b><br/> <b><a href="#">André Pereira</a></b>, Faculdade de Direito da Universidade de Coimbra</p> |
| <b>11:00</b> | <b>Coffee Break / Poster Viewing</b>  |
| <b>11:30</b> | <p><b>Genome Editing with Programmable Nucleases</b><br/> <b><a href="#">Dana Carroll</a></b>, University of Utah School of Medicine, Salt Lake City, UT, USA</p>   |
| <b>12:15</b> | <b>Basic and Clinical Research Awards Ceremony</b>  |
| <b>12:45</b> | <b>SPGH Award Conference</b>  |
| <b>12:55</b> | <b>Closing Session</b>  |