



20^a REUNIÃO ANUAL 2016

Fundação Bissaya Barreto COIMBRA 10-12 de NOVEMBRO 2016

Scientific programme

Day 1 Thursday, 10 November

09.30 h **Registration Opening**

10.00 h **Clubs (Parallel Sessions)**

- **Cytogenetic and Molecular Genetics Club**

Medical Genetics and Clinical Dysmorphology Club

14.15 h - **Opening & Welcome Session**

Joana Barbosa de Melo, SPGH President
Sérgio Bernardo Sousa, SPGH Secretary

14.30 h – **Neurogenetics**

Novel Therapeutic approaches – Machado Joseph disease

Luis Almeida, CNC, Faculty of Pharmacy, Coimbra, Portugal (definitive title to be announced)

Genomics in neurodegenerative diseases

Rita Guerreiro and José Braz, London, UK (definitive title to be announced)

15.30 h- **Selected Clinical Cases Session I**

16.00 h - **Coffee Break / Poster Viewing**

16.25 h – **Corporate Symposium (Illumina)** – title to be announced

16.45 h – **Genome structure and phenotype: 16p11.2 rearrangements as an example**

Alexandre Reymond, Un. Lausanne, Switzerland

17.15 h - **Selected Oral Communications I**

18.45 h - **SPGH Assembly**



Day 2 Friday, 11 November

09.00 h – **Selected Oral Communications II**

10.30 h – **The non-coding Genome – breaking TADs**

Stefan Mundlos, Max Plank Institute, Berlin, Germany (definitive title to be announced)

11.15 h - **Coffee Break / Poster Viewing and discussion**

12.00 h - **Oncogenetics**

Thierry Frebourg, Rouen University Hospital, Rouen, France (definitive title to be announced)

Paula Soares, Ipatimup, Porto, Portugal (definitive title to be announced)

13.00 h - **Lunch**

14.15 h - **Challenges in interpreting new variants**

Clinical Interpretation of Genetic Variants – Recommendations

Steven Harrison, Harvard University, USA -

Encarna Gomez, Maastricht, The Netherlands (title to be announced)

Leslie Biesecker, National Human Genome Research Institute, USA (definitive title to be announced)

José Carlos Machado, IPATIMUP, Portugal (definitive title to be announced)

16.15 h - **Coffee Break / Poster Viewing**

16.40 h – **Corporate Symposium (Sophia Genetics)** – title to be announced

17.00 h – **Prenatal Diagnosis: where are we going?**

Isabel Marques Carreira, FMUC, Coimbra, Portugal

Fabiana Ramos, HP-CHUC, Coimbra, Portugal

17.45 h– **Public Policy “Centros de Referência em Portugal – Estratégia para as Doenças Genéticas”**

Jorge Sequeiros, IPATIMUP, Porto, Portugal

João Lavinha, INSA, Lisboa, Portugal

20.00 h - **Conference Dinner**



Day 3 Saturday, 12 November

09.00 h- **Selected Clinical Cases Session II**

09.30 h – **Bioethics debate: Carrier Screening**

Heloísa Santos, SPGH, Lisboa, Portugal

Lidewij Henneman, VU University Medical Center, Amsterdam, The Netherlands

10.30 h – **Sessão Comemorativa do 20º aniversário da SPGH**

10.45 h - **Coffee Break / Poster Viewing**

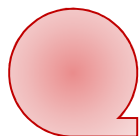
11.15 h – **Mosaicism and Disease**

Leslie Biesecker, National Human Genome Research Institute, USA (definitive title to be announced)

12.15 h - **SPGH 2015 Award Conference – to be announced**

12.45 h - **Basic and Clinical Research Awards Ceremony**

12.55 h - **Closing Session**



IMPORTANT DATES :

- ✓ **Deadline for *Abstract* Submission: 10.10. 2016**
- ✓ **Deadline for *Reduced Price* Registration: 27 .10. 2016**