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 – Mosaicim 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