



## Preliminary Programme

### Day 1 Thursday, November 5

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#### Clubs (Parallel Sessions)

- 14:00 **Registration Opening**
- 14:30 **Human Variome Project Meeting (UNESCO)**  
*Chairs: Maria Luís Cardoso, Rosário Santos (CGMJM)*
- 16:30 **Cytogenetic and Molecular Genetics Club**  
*Chairs: Sofia Dória (FMUP), Rosário Santos (CGMJM)*
- 16:30 **Medical Genetics and Clinical Dismorphology Club**  
*Chairs: Ana Fortuna (CGMJM), Gabriela Soares (CGMJM)*

### Day 2 Friday, November 6

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- 08:30 **Registration Opening**
- 09:00 **Opening & Welcome Session**  
*Margarida Reis Lima, Susana Fernandes, Cecília Correia*
- 09:15 **Neuromuscular Disorders**
- Genetic variant databases: their present and future roles in Human Genetics**  
*Jorge Oliveira (Porto, CGMJM/CHP)*
- Advances in diagnosis and treatment of Duchenne muscular dystrophy**  
*Kevin Flanigan (Columbus, USA, Nationwide Children's Hospital)*
- 10:30 **Personal genomics, horoscopes and forensic genetics**  
*António Amorim (Porto, IPATIMUP)*
- 11:00 **Coffee Break / Poster Viewing**
- 11:30 **Pre-clinical studies for the development of pharmacological treatment in hereditary amyloidosis**  
*Maria João Saraiva (Porto, IBMC)*
- 12:00 **Oral Communications I**



# 19ª REUNIÃO ANUAL

ORDEM DOS MÉDICOS - PORTO 5 - 7 NOVEMBRO 2015

SPGH Sociedade Portuguesa de Genética Humana

- 13:00 Lunch / Poster Viewing**
- 14:30 Prenatal Diagnosis**
- aCGH in PND: data, decisions and dilemmas**  
*Paula Rendeiro (Porto, CGC Genetics)*
  - NIPT workflow implementation and technology considerations**  
*Francesco Fiorentino (Roma, IT, GenomaLaboratory)*
- 15:30 Face2Gene: facial dysmorphology automated analysis in clinical genetics**  
*Nicole Fleischer (NYC, USA, FDNA)*
- 16:00 Coffee Break / Poster Viewing**
- 16:30 From genes to genomes in Medical Genetics: new possibilities and new challenges**  
*Joris Veltman (Nijmegen, NL, RadboudUMC)*
- 17:30 SPGH Assembly**
- 20:30 Conference Dinner**

## Day 3 Saturday, November 7

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- 08:45 Oral Communications II**
- 10:00 News in diagnosis and treatment of Lysosomal Storage Disorders (LSD)**  
*Paula Garcia (Coimbra, CHUC)*
- 10:30 The future of Human Genetics training in Europe**  
*Feliciano Ramos-Fuentes - President, ESHG (Zaragoza, ES, H.Clinico Universitario)*
- 11:00 Coffee Break / Poster Viewing**
- 11:30 Oncogenetics**
- Cancer Immunotherapy and Tumor Escape**  
*Haaken Norell (Lisboa, IMM)*
  - The changing landscape in BRCA1/2 testing for ovarian cancer predisposition and treatment: is tumor testing first the new standard practice ?**  
*Nicoline Hoogerbrugge (Nijmegen, NL, RadboudUMC)*



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13:00 **Lunch / Poster Viewing**

14:00 **De novo mutations in Intellectual Disability**  
*Joris Veltman* (Nijmegen, NL, RadboudUMC)

14:45 **GenEthics: Bioethics Committee Session**

**Bioethical evaluation of the new reproductive techniques for mitochondrial disorders prevention**

**Impact and clinical spectrum of mitochondrial disorders**  
*Isabel Santana* (Coimbra, CHUC)

**Mitochondrial disorders prevention - the new reproductive medicine techniques and its dilemmas**  
*Célia Ventura* (Lisboa, INSA)

**Ethical considerations and recommendations**  
*Heloísa G.Santos* (Lisboa, President of SPGH Ethics Committee)

15:45 **SPGH 2015 Award Conference**

16:15 **Basic and Clinical Research Awards Ceremony**

16:30 **Closing Session**



## DEADLINES:

- ✓ **Abstract Submission: October 12**
- ✓ **Reduced Price Registration: October 26**

## INFORMAÇÃO

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