



# 19<sup>a</sup> REUNIÃO ANUAL da SOCIEDADE PORTUGUESA de GENÉTICA HUMANA

## COMISSÃO ORGANIZADORA

Margarida Reis Lima | Susana Fernandes | Cecília Correia

## COMISSÃO CIENTÍFICA

Ana Berta Sousa | Carla Oliveira | Filipa Carvalho  
Joana Melo | Jorge Saraiva | Lina Ramos  
Luísa Romão | Manuel Teixeira | Paula Faustino  
Sofia Dória | Susana Fernandes

## INFORMAÇÃO & CONTACTOS

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## PATROCINADORES

DIAMANTE



PLATINA



OURO



PRATA



## PARCEIROS CIENTÍFICOS & INSTITUCIONAIS



# PORTO

5 - 7 NOVEMBRO 2015



Ordem dos Médicos  
Rua Delfim Maia, 405  
4200-256 Porto, Portugal  
[www.nortemedico.pt](http://www.nortemedico.pt)





## Friday, November 6

- 08:30 **Registration Opening**
- 09:00 **Opening & Welcome Session**  
*Margarida Reis Lima, Susana Fernandes, Cecília Correia*
- 09:15 **Neuromuscular Disorders**  
*Chairs: Ana Fortuna, Rosário Santos*
- Genetic variant databases: their present and future roles in Human Genetics**  
*Jorge Oliveira (Porto, CGMJM/CHP)*
- Advances in the diagnosis and treatment of Duchenne muscular dystrophy**  
*Kevin Flanigan (Columbus, USA, Nationwide Children's Hosp.)*
- 10:30 **Personal genomics, horoscopes and forensic genetics**  
*Chair: Sérgio Castedo*  
*António Amorim (Porto, IPATIMUP)*
- 11:00 **Coffee Break / Poster Viewing**
- 11:30 **Pre-clinical studies for the development of pharmacological treatment in hereditary amyloidosis**  
*Chairs: Hildeberto Correia, Juliette Dupont*  
*Maria João Saraiva (Porto, IBMC)*
- 12:00 **Oral Communications I**  
*Chairs: Lina Ramos, Sofia Dória*
- 13:00 Lunch / Poster Viewing**
- 14:30 **Prenatal Diagnosis**  
*Chairs: Filipa Carvalho, Isabel Carreira*
- aCGH in PND: data, decisions and dilemmas**  
*Paula Rendeiro (Porto, CGC Genetics)*
- NIPT workflow implementation and technology considerations**  
*Marina Baldi (Roma, IT, GenomaLaboratory)*
- 15:30 **Face2Gene: facial dysmorphology automated analysis in clinical genetics**  
*Chair: Miguel Rocha*  
*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**  
*Chairs: Jorge Pinto-Basto, João Silva*  
*Joris Veltman (Nijmegen, NL, RadboudUMC)*
- 17:30 **SPGH Assembly**
- 20:30 Conference Dinner**

## Saturday, November 7

- 08:45 **Oral Communications II**  
*Chairs: Ana Berta Sousa, Luísa Romão*
- 10:00 **News in diagnosis and treatment of Lysosomal Storage Disorders (LSD)**  
*Chairs: João Paulo Oliveira, Carolino Monteiro*  
*Paula Garcia (Coimbra, CHUC)*
- 10:30 **The future of Human Genetics training in Europe**  
*Chairs: Margarida Reis Lima, Joana Melo*  
*Feliciano Ramos-Fuentes - President, ESHG (Zaragoza, ES, H.Clinico Universitario)*
- 11:00 **Coffee Break / Poster Viewing**
- 11:30 **Oncogenetics**  
*Chairs: Manuel Teixeira, Carla Oliveira*
- 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)*
- 13:00 Lunch / Poster Viewing**
- 14:00 **De novo mutations in Intellectual Disability**  
*Chairs: Patrícia Maciel, Sérgio B. Sousa*  
*Joris Veltman (Nijmegen, NL, RadboudUMC)*
- 14:45 **GenEthics: Bioethics Committee Session**
- Bioethical evaluation of the new reproductive techniques for mitochondrial disorders prevention**  
*Chairs: Jorge Sequeiros, Heloísa G.Santos*
- 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**



## Thursday, November 5

### 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 Dysmorphology Club**  
*Chairs: Ana Fortuna (CGMJM), Gabriela Soares (CGMJM)*