



SPGH

Sociedade Portuguesa de Genética Humana



## 22<sup>a</sup> Reunião Anual / 22<sup>nd</sup> Annual Meeting

15 - 17 November 2018

### Preliminary Programme\_28<sup>th</sup> August

#### 15<sup>th</sup> Nov

09:00h **Registration**

10:00h **SPGH Club meetings (concurrent)**

- Molecular and Cytogenetics Club
- Medical and Clinical Dysmorphology Club

12:30h **ERNs in Portugal**

Panel Discussion (Current representatives in Portugal)

14:00h **Opening & Welcome**

14:15h **Keynote Lecture:**

*(The Genetics of Intellectual Disability - Exomes & beyond)*

**Anita Rauch** (Institute of Medical Genetics, Zurich)

15:00h **Epigenetics**

- *The role of Imprinting in Cancer*  
**David Monk** (Inst. d'Investigació Biomédica, Barcelona)
- *Imprinting errors in spermatogenic cells of infertile patients*  
**Joana Marques** (Dept. Patologia, HSJ, Porto)
- *Establishing the human epigenome in development and pluripotency*  
**Peter Rugg-Gunn** (Babraham Institute, Univ. Cambridge)

16:30h **Coffee-break / poster viewing**

16:45h **Poster discussions**

17:10h **Corporate symposium** (Blueprint Genetics)

- (tba)

17:30h **Selected Oral Presentations (I)**

18:30h **SPGH General Assembly**

**16<sup>th</sup> Nov**

08:45h **Selected Oral Presentations (II)**

09:45h **Meet the expert: Update on Batten's Disease** (Sponsored by BioMarin)

- *An algorithm for CLN2 Diagnosis*  
**Miguel Leão** (Dept<sup>o</sup> Genética Humana, HSJ, Porto)
- *From early diagnosis to early intervention*  
**Paul Gissen** (UCL Great Ormond Street Hospital for Children, London)

10:30h **Coffee-break / poster viewing**

10:45h **Poster discussions**

11:15h **Immunotherapy & Genetics**

- *Harnessing the immunotherapy revolution for the treatment of colorectal cancer: neo-antigens and beyond*  
**Noel de Miranda** (LUMC, Leiden)
- *The genetic basis of the response to Cancer Immunotherapies* (Sponsored by AstraZeneca)  
**Vassiliki Kotoula** (AUTH School of Medicine, Thessaloniki)
- *CAR-T for the treatment of T cell malignancies*  
**John F. DiPersio** (Alvin J. Siteman Cancer Center & Div. of Oncology, WU Medical School)

13:00h **Lunch break**

14:00h **Neurogenetics & Therapy**

- *Advanced therapies in SMA: beyond the clinical trials*  
**Eduardo Tizzano** (H. Valle Hebron, Barcelona)
- *(Therapeutic developments in Huntington's and other neurodegenerative disorders)*  
**Bernhard Landwehrmeyer** (Dept. Neurology, Univ. Ulm)
- *LRP10: a novel player in late-onset inherited synucleinopathies*  
**Wim Mandemakers** (Erasmus Medical Center, Rotterdam)
- *(Biomarkers in Alzheimer's disease)*  
**Luís Maia** (Dept<sup>o</sup>. Neurologia, CHP, Porto)

16.15h **Coffee-break / poster viewing**

16:30h **Poster discussions**

17:00h **Corporate symposium** (ILC Lda.)

- *Single molecule sequencing - 10xGenomics* (tba)

17:30h **Keynote Lecture:**

- *(Big data and machine learning in genomics)* (tbc)

20:00h **Congress Dinner**

**17<sup>th</sup> Nov**

09:00h **Selected Clinical Case Reports**

10:00h **Ethical Issues in Genome Editing**  
Panel Discussion (SPGH Bioethics Committee)

11:00h **Coffee-break**

11:30h **Keynote Lecture:**

*Unlocking the diagnostic potential for circulating DNA*

**Dennis Lo** (Faculty of Medicine, The Chinese University of Hong Kong)

12:15h **SPGH Award Lecture**

12:30h **SPGH Awards Cerimony**

12:45h **Closing Session**

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14:30h **SPGH closed meetings** (By invitation only)

- SPGH Committee of Public Policies & Education in Genetics  
(Together with ESHG Education Committee)
- Genome.pt network