



Preliminary Programme_17 Oct

15th Nov

09:00h **Registration**

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

- Molecular and Cytogenetics Club
- Medical and Clinical Dysmorphology Club

12:30h **Satellite Meeting: European Reference Networks in Portugal**

Panel Discussion (Current representatives of ERNs in Portugal)

14:00h **Opening & Welcome**

14:15h **Keynote Lecture:**

- *The Genetic Landscape of ID*
Anita Rauch (Institute of Medical Genetics, Zurich)

15:00h **Epigenetics**

- *The role of Imprinting in Cancer*
David Monk (Inst. d'Investigació Biomèdica, 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**

17:15h **Selected Oral Presentations (I)**

18:15h **SPGH General Assembly**

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

09:45h **Meet the expert: CLN2, a form of Batten Disease** (Sponsored by BioMarin)

- *An algorithm for CLN2 Diagnosis*
Miguel Leão (Dept^o Genética Humana, HSJ, Porto)
- *From early diagnosis to early intervention*
Angela Schulz (Department of Paediatrics, University Medical Center Hamburg-Eppendorf, Hamburg)

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 response to checkpoint inhibitors*
Vassiliki Kotoula (AUTH School of Medicine, Thessaloniki) (Sponsored by AstraZeneca)
- *CAR-T for the treatment of T cell malignancies*
John F. DiPersio (Alvin J. Siteman Cancer Center / Div. Oncology, Washington Univ. Medical School)
(Sponsored by FLAD – *Luso-American Development Foundation*)

13:00h **Lunch break**

14:00h **Neurogenetics & Therapy**

- *Towards modifying disease progression and onset in Huntington's Disease and other genetic neurodegenerative disorders: hopes and challenges.*
Bernhard Landwehrmeyer (Dept. Neurology, Univ. Ulm)
- *LRP10: a novel player in late-onset inherited synucleinopathies*
Wim Mandemakers (Erasmus Medical Center, Rotterdam)
- *Fluid Biomarkers in Neurodegenerative Diseases*
Luís Maia (Dept^o. Neurologia, CHP, Porto)
- *Advanced therapies in SMA: beyond the clinical trials*
Eduardo Tizzano (H. Valle Hebron, Barcelona) (Sponsored by Biogen)

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

16:30h **Poster discussions**

16:45h **Corporate symposia**

- *A clinical case: From the sample to the analysis on the NGS Workflow*
Alba Mota; Ivan Lesende (SoQuímica /Agilent)
- *10x Genomics technology in Human Genetics: from bulk long range sequencing to Copy Number variation in Single Cells*
Hannes Arnold (10X Genomics, Germany) (ILC Lda.)

17:45h **Keynote Lecture:**

- *The genetics of cognitive epigenetics*
Hans van Bokhoven (Radboud U. Medical Center / Donders Institute for Brain, Cognition & Behaviour, Nijmegen)

20:00h **Congress Dinner**

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

10:00h **BIOETHICS DEBATE: Genome editing – what, why and how? The ethical issues**
(SPGH Bioethics Committee)

- *CRISPR-Cas 9 – Method and potential applications*
André Travessa (S. Genética, HSM)
- *Genome editing - from benefits to ethical limits*
Heloísa Santos (SPGH Bioethics Committee / S. Genética, HSM, FMUL)
- *Current Portuguese legislation and ethical guidelines*
André Pereira (SPGH Bioethics Committee / Fac. of Law, Univ. Coimbra)

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**

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

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