



23^a Reunião Anual / 23rd Annual Meeting

14 - 16 November 2019

Preliminary Programme

Thursday, 14th Nov

09h00-09h45 **Registration**

09h45-11h45 **SPGH Club meetings (concurrent)**

➤ **CLUB 1: Cytogenetic and Molecular Genetics Club**

Chairs: Joana Barbosa Melo and Celeste Bento

➤ **CLUB 2: Dysmorphology and Clinical Genetics Club**

Chairs: Jorge Saraiva and Sérgio Sousa

11h45-12h45 **Workshop:** European Reference Networks, what is in there for me?

Chairs: Carla Oliveira and Lina Ramos

11h45-11h55 **Brief ERNs introduction**

➤ **Sérgio Sousa**, (*Centro Hospitalar e Universitário de Coimbra-CHUC, Portugal*)

11h55-12h10 **Selected ERN: EpiCARE**

EpiCARE for rare and complex epilepsies - overall organization and what is in there for genetics professionals?

➤ **Gaetan Lesca**, (*MCU-PH chez Hospices Civils de Lyon, France*)

12h10-12h20 **Questions and Answers**

12h20-12h30 EpiCARE in Portugal.

➤ **Francisco Sales**, (*Centro Hospitalar e Universitário de Coimbra-CHUC, Portugal*)

12h30-12h45 **General discussion**

13h00-13h45 **Symposium (to be announced)**

14H00-14h15 Opening & Welcome

14h15-14h50 Keynote Lecture 1: “Implementing whole-genome sequencing in routine - consequences to the NHS”

➤ *Helen Firth, (Cambridge University Hospitals, UK)*

14h50-16h20 Invited Symposium I (IS-I): “Therapy for Genetic Diseases”

14h50-15h20 ISI-1: “Luxturna, First Gene Therapy for Inherited Blindness associated with the RPE65 gene”

➤ *(Speaker to be confirmed)*

15h20-15h50 ISI-2: “From disease mechanism to new treatments in X-linked hypophosphatemia”

➤ *Helena Gil Peña, (Hospital Universitario Central de Asturias, Oviedo, Spain)*

15h50-16h20 ISI-3: “Targeted therapy in patients with PIK3CA-related overgrowth syndrome”

➤ *Guillaume Canaud, (Necker, Paris, France)*

16h20-17h00 Coffee-break / Poster viewing and Discussion

17h00-18h00 5 Selected Oral Presentations I (Basic Research)

18h00 SPGH General Assembly

Friday 15th Nov

09h00-10h00 5 Selected Oral Presentations II (Clinical Research)

10h00-11h00 Panel Discussion I (PDI): “SPGH and SPO-Portuguese Society of Oncology: Building bridges”

10h00-10h20 PDI-1: “Somatic testing of *BRCA* genes in tumours: implications for germline testing: The Oncologist perspective”

➤ *Gabriela Sousa, (Portuguese Institute of Oncology (IPO), Coimbra, Portugal)*

10h20-10h40 PDI-2: “Somatic testing of *BRCA* genes in tumours: implications for germline testing: The Geneticist perspective”

➤ *Ana Berta Sousa, (Hosp. Santa Maria, Lisboa, Portugal)*

10h40-11h00 Discussion

11h00-11h30 Coffee-break / Poster Viewing and Discussion

- 11h30-13h00 Invited Symposium II (ISII) “Mosaicism: Old but Hot topic”**
- 11h30-12h00 **ISII-1:** “Mosaicism in human blastocysts: incidence, prevalence and diagnostic capabilities in PGT-A cycles”
➤ **Antonio Capalbo**, (Igenomix, Rome, Italy)
- 12h00-12h30 **ISII-2:** “Mosaicism in cancer”
➤ **Stefan Aretz**, (University of Bonn, Germany)
- 12h30-13h00 **ISII-3:** “Parental Mosaicism in “De Novo” Epileptic Encephalopathies”
➤ **Stephanie Baulac**, (Hôpital Pitié-Salpêtrière, Paris, France)

13h00-14h00 Lunch break

14h00-15h00 Poster Viewing and Discussion

15h00-16h30 Invited Symposium III (ISIII): “Understanding disease through Big Data”

- 15h00-15h30 **ISIII-1:** “Systems-level transcriptional regulation and its perturbation in colorectal cancer”
➤ **Nuno Barbosa Morais**, (IMM, Lisbon, Portugal)
- 15h30-16h00 **ISIII-2:** “Sequence variant effect predictions of single nucleotide variants in model organisms: a computational approach”
➤ **Pedro Beltrão**, (Sanger Centre, Cambridge, UK)
- 16h00-16h30 **ISIII-3:** “Big data in healthcare in Rare genetic diseases”
➤ **Ignacio Medrano**, (Ramón y Cajal Hospital, Madrid, Spain)

16h30-16h45 Coffee-break

16h45-17h45 Invited Symposium IV (ISIV): “The importance of Small and Large: Single cells and whole Genomes”

- 16h45-17h15 **ISIV-1:** “Single-cell sequencing and its importance for human genetics”
➤ **Malte Spielmann**, (Max Planck Institute Berlin, Germany)
- 17h15-17h45 **ISIV-2:** “Delineating the structure of chromosome rearrangements using multiple WGS technologies”
➤ **Anna Lindstrand**, (Karolinska Institute, Stockholm, Sweden)

17h45-19h05 Panel Discussion (PDII): “National and International Consortia in Human Genetics”

- 17h45- 18h05 **PDII-1:** “The experience from the UK – Lessons from national projects: DECIPHER, DDD and 100,000 Genomes Project”

- *Helen Firth* (Cambridge University Hospitals, UK)
- 18h05-18h25 **PDII-2: “The GenomePT Consortium”**
 - *Manuel Santos*, (iBIMED, Aveiro, Portugal)
- 18h25-18h45 **PDII-3: “1 Million Genomes project”**
 - *Astrid Vicente*, (INSA, Lisbon, Portugal)
- 18h45-19h05 **Open discussion with the audience**

- 20h30 Gala Dinner**

Saturday 16th Nov

- 08h45-09h45 8 Selected Oral Presentations III (Clinical cases)**

- 09h45-10h45 BioEthics Debate: “title to be announced”**
Chair: Heloísa Santos

- 10h45-11h30 Coffee-break / Poster Viewing**

- 11h30-12h20 Keynote Lecture 2: “Functional characterization and therapeutic targeting of gene regulatory elements”**
 - *Nadav Ahituv*, (UCSF, USA)

- 12h20-12h50 SPGH Award Lecture**
- 12h50-13h15 SPGH Awards Ceremony**
- 13h15-13h30 Closing Session**