



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:** “Brain somatic mutations in focal malformations of cortical development with epilepsy”

➤ **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 Consortiums 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: “BIG Genomic DATA - Bigger scientific advantages means bigger ethical responsibilities”

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