



23ª Reunião Anual / 23rd Annual Meeting

14 - 16 November 2019

Programme

Thursday, 14th Nov

09h00-09h45 Registration

09h45-11h45 SPGH Club meetings (concurrent)

CLUB 1: *Cytogenetic and Molecular Genetics Club*

Chairs: Joana Barbosa de Melo and Celeste Bento

CLUB 2: *Dysmorphology and Clinical Genetics Club - Genetic syndromes and the mTOR/PIK3CA pathway*

Chairs: Jorge Saraiva and Sérgio B. 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 B. Sousa (*Centro Hospitalar e Universitário de Coimbra-CHUC, Portugal*)

11h55-12h10 Selected ERN: EpiCARE

EpiCARE-ERN for rare and complex epilepsies: overall organization and what is in there for the geneticist

Gaetan Lesca (*Lyon University Hospital, France, 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

12h45-13h15 Lunch box

13h15-13h45 Corporate Symposium - SOPHiA GENETICS: Molecular Diagnosis of Neurometabolic Diseases Using SOPHiA Clinical Exome Solution

Sofia Isabel Gouveia (*Hospital Clínico Universidad de Santiago de Compostela, Spain*)

14H00-14h15 Opening & Welcome

Isabel Carreira, Eunice Matoso, Rosário Pinto Leite, Conselho Diretivo da FMUC

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

Lucy Raymond (*Cambridge Institute for Medical Research, UK*)

Chairs: Carla Oliveira

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

Chairs: Sérgio B. Sousa and Ana Berta Sousa

14h50-15h20 ISI-1: “From gene discovery to gene therapy in 20 years: the choroideremia case”

Miguel Seabra (*Centro de Estudos de Doenças Crónicas - CEDOC, Nova Medical School, Lisbon, Portugal*)

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 Hospital, Paris, France*)

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

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

Chairs: Susana Fernandes and João Gonçalves

18h00 SPGH General Assembly

Chairs: Isabel Carreira, Eunice Matoso, Rosário Pinto Leite

Friday 15th Nov

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

Chairs: Sérgio B. Sousa and Luísa Romão

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

Chairs: Sofia Maia and Sara Meireles

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 (*Hospital 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”

Chairs: Joana Barbosa de Melo and Lina Ramos

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: “Somatic Mosaicism in Tumour Genetics”

Stefan Aretz (*University of Bonn, Germany*)

12h30-13h00 ISII-3: “Brain somatic mutations in focal malformations of cortical development with epilepsy”

Stéphanie Baulac (*Institut du Cerveau et de la Moelle Épinrière, Paris, France*)

13h00-14h00 Lunch break

14h00-15h00 Poster Viewing and Discussion

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

Chairs: Rosário Santos and Carolino Monteiro

15h00-15h30 ISIII-1: “Tumour transcriptomes reveal a prognostic alternative splicing signature in colorectal cancer”

Nuno Barbosa Morais (*Instituto de Medicina Molecular, FMUL, Lisbon, Portugal*)

15h30-16h00 ISIII-2: “Network based prioritization of genetically associated genes for 1225 human traits”

Pedro Beltrão (*EMBL-EBI, 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”

Chairs: Sofia Dória and João Gonçalves

16h45-17h15 ISIV-1: “Single-cell sequencing and its importance for human genetics”

Malte Spielmann (*Max Planck Institute for Molecular Genetics, 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”

Chairs: Carla Oliveira and Jorge Saraiva

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

Lucy Raymond (*Cambridge Institute for Medical Research, UK*)

18h05-18h25 PDII-2: “GenomePT: From Gene Panels to WES, WGS and Population Genomics”

Manuel Santos (*Institute of Biomedicine, University of Aveiro, Portugal*)

18h25-18h45 PDII-3: “1 Million Genomes Project”

Astrid Vicente (*Instituto Nacional de Saúde Doutor Ricardo Jorge, Lisbon, Portugal*)

18h45-19h05 Open discussion with the audience

20h30 Conference Dinner

Saturday 16th Nov

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

Chairs: Ana Berta Sousa and João Silva

09h45-10h45 BioEthics Debate (BE): “BIG Genomic DATA - Bigger scientific advantages means bigger ethical responsibilities”

Chairs: Heloisa Santos and André Pereira

BE-1: International genomic databases - Historical background. From Iceland to Africa.

Célia Ventura (Instituto Nacional de Saúde Doutor Ricardo Jorge, Lisbon, Portugal)

BE-2: Cross-border flow of human genetic data - The portuguese laws and norms. New law of National Health Service.

Cristina Caldeira (Universidade Nova de Lisboa, Lisboa, Portugal)

BE-3: National genomic databases – Ethical considerations including importance of transparency and other ethical standards. European Digital Single Market Big Data.

Carolino Monteiro (Faculdade de Farmácia da Universidade de Lisboa, Lisboa, Portugal)

Conclusions and general discussion

Heloisa Santos (President of SPGH's Committee of Ethics, Portugal)

10h45-11h30 Coffee-break

11h30-12h20 Keynote Lecture 2: “Functional characterization and therapeutic targeting of gene regulatory elements”

Nadav Ahituv (University of California San Francisco , California, USA)

Chairs: Isabel Carreira and President Elected of SPGH 2021

12h20-12h50 SPGH Award Lecture

Chairs: Carla Oliveira and João Gonçalves

12h50-13h15 SPGH Awards Ceremony

Chairs: Isabel Carreira and Carla Oliveira

13h15-13h30 Closing Session

Chairs: Isabel Carreira, Eunice Matoso, Rosário Pinto Leite