SPGH Sociedade Portuguesa de Genética Humana

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

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

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

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"

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 Astúrias, Oviedo, Spain)*

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

Guillaume Canaud *(Necker, Paris, France)*

Chairs: Sérgio B. Sousa and Ana Berta Sousa

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"

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

Chairs: Sofia Maia and Sara Meireles

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 Tumour Genetics"

Stefan Aretz *(University of Bonn, Germany)*

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

Stephanie Baulac *(Institut du Cerveau et de la Moelle Épinière , Paris, France)*

Chairs: Joana Barbosa de Melo and Lina Ramos

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: "Tumour transcriptomes reveal a prognostic alternative splicing signature in colorectal cancer"
Nuno Barbosa Morais *(IMM, Lisbon, Portugal)*

15h30-16h00 ISIII-2: "Network based prioritization of genetically associated genes for 1225 human traits"
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)*
Chairs: Rosário Santos and Carolino Monteiro

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)*
Chairs: Sofia Dória and João Gonçalves

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

18h05-18h25 PDII-2: "GenomePT: From Gene Panels to WES, WGS and Population Genomics"
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
Chairs: Carla Oliveira and Jorge Saraiva

20h30 Gala 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"

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

Célia Ventura *(INSA, 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

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

Chairs: Heloísa Santos and André Pereira

10h45-11h30 Coffee-break

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

Nadav Ahituv *(UCSF, 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

CLUBE DE CITOGENÉTICA E GENÉTICA MOLECULAR
Sociedade Portuguesa de Genética Humana
14 DE NOVEMBRO DE 2019

COIMBRA, FUNDAÇÃO BISSAYA BARRETO
Organização: Celeste Bento e Joana Barbosa de Melo

9h45m – Apresentação e discussão de casos/dilemas

"Holoprosencefalia por interrupção do gene *SHH* em t(7;17)"
Raquel Rodrigues, Márcia Rodrigues, Marta P. Soares, Rosário Silveira-Santos, Ana Sousa, Ana Berta Sousa. Laboratório de Genética - Serviço de Genética Médica, Departamento de Pediatria, Hospital de Santa Maria, Centro Hospitalar Universitário de Lisboa Norte EPE, Centro Académico de Medicina de Lisboa, Lisboa

"Uma gestação, dois desequilíbrios, três progenitores"
Marta C. Pinto, Alexandra Mascarenhas, Nuno Lavoura, Isabel Marques Carreira, Joana Barbosa de Melo. Laboratório de Citogenética e Genómica, Faculdade de Medicina da Universidade de Coimbra, Coimbra

"O desafio das novas tecnologias de sequenciação no diagnóstico de doenças genéticas raras - um caso de síndrome de Burn-McKeown"
João Parente Freixo, Sofia Nunes, Paulo Silva, Jorge Oliveira, Jorge Sequeiros. Centro de Genética Preditiva e Preventiva, IBMC, Porto

"Relevance of familial studies when analyzing large gene panels."
Sofia Quental. I3S/IPATIMUP, Universidade do Porto, Porto

"Co-occurrence of a moderate effect *TP53* mutation with additional cancer related variants."
Sónia Sousa. I3S/IPATIMUP, Universidade do Porto, Porto

10.45m – Painel de discussão "*Laboratórios de Genética Clínica Laboratorial – Onde estamos? Qual o futuro*?"
Moderadores: Celeste Bento, Joana Barbosa de Melo, Rosário Pinto-Leite, Sofia Dória, Sofia Quental

11h45m - Fim

SPHG, Coimbra 2019

Thursday, November 14, 2019, 09:45 hrs

Club Meeting:	Dysmorphology and Clinical Genetics Club
Date:	Thursday, November 14, 2019
Time:	09:45-11:45 hrs
Room:	--------
Co-Organiser(s):	Jorge Saraiva 20741@chuc.min-saude.pt Sérgio Sousa 8911@chuc.min-saude.pt

PROGRAMME OVERVIEW:

A. TOPIC AND AIM OF THE CLUB

In this session participants will be reminded of new developments in syndromes associated to the mTOR/PIK3CA pathway and patients with diagnosis of this spectrum and other known and unknown diagnosis will be discussed.

B. CONTENT OF THE SESSION

Each presentation will be of 5 minutes per Genetics Unit in all sessions but the first.

09:45 – Welcome – **Jorge Saraiva e Sérgio Sousa**

09:45 – 10:15
Genetic syndromes and the mTOR/PIK3CA pathway
Presenters Mário Noro and Sara Ribeiro

10:15 – 10:45
Patients with syndromes associated to the mTOR/PIK3CA pathway
Presenters – GenetIc Units of Centro Hospitalar Universitário de São João, Centro Hospitalar Universitário do Porto, Centro Hospitalar e Universitário de Coimbra, Centro Hospitalar Universitário Lisboa Central and Centro Hospitalar Universitário de Lisboa Norte

10:45 – 11:00
Other patients with a known diagnosis
Presenters - GenetIc Units, Centro Hospitalar Universitário de São João, Centro Hospitalar Universitário do Porto, Centro Hospitalar e Universitário de Coimbra, Centro Hospitalar Universitário Lisboa Central and Centro Hospitalar Universitário de Lisboa Norte

11:00 – 11:30
Patients without an established diagnosis
Presenters - GenetIc Units, Centro Hospitalar Universitário de São João, Centro Hospitalar Universitário do Porto, Centro Hospitalar e Universitário de Coimbra, Centro Hospitalar Universitário Lisboa Central and Centro Hospitalar Universitário de Lisboa Norte

11:30 – Session closing, **Jorge Saraiva e Sérgio Sousa**

C. SESSION LEARNING OUTCOME

Attendees will learn about:

1. New developments in syndromes associated to the mTOR/PIK3CA pathway
2. Diagnosis of other syndromes
3. How to try to establish a diagnosis

D. SHORT DESCRIPTION OF THE INTERACTIVE ELEMENT OF THE SESSION

- Club meeting Speakers will have open questions at the end of their presentations that will be discussed in up to 5 minutes with the audience after each one.

- In case the discussion with the audience becomes vague, Workshop Chairs will stimulate the Speakers to raise provocative issues for discussion.

- Delegates will be stimulated to engage, interact and provide feedback by the Chairs of the Clubs as well as by the Speakers of the session.

E. LIST OF WORKSHOP SPEAKERS

Name of participant	Affiliation	Email
Mário Noro	Medical Genetics Unit, Hospital Pediátrico, Centro Hospitalar e Universitário de Coimbra	11365@chuc.min-saude.pt
Sara Ribeiro	Medical Genetics Unit, Hospital Pediátrico, Centro Hospitalar e Universitário de Coimbra	11197@chuc.min-saude.pt
Genetics Unit	Centro Hospitalar Universitário de São João	
Genetics Unit	Centro Hospitalar Universitário do Porto	
Genetics Unit	Hospital Pediátrico, Centro Hospitalar e Universitário de Coimbra	
Genetics Unit	Centro Hospitalar Universitário Lisboa Central e Centro Hospitalar Universitário de Lisboa Norte	
Genetics Unit	Centro Hospitalar Universitário Lisboa Central e Centro Hospitalar Universitário de Lisboa Norte	

F. WORKSHOP TECHNICAL REQUIREMENTS

- WIFI in the computer from the room
- One large screen for speakers presentations
- Slide upload in the room
- Four sitting places in the podium (1/2 speakers and 2 chairs)