26TH ANNUAL MEETING

Sociedade Portuguesa de Genética Humana

2022 NOV 17TH - 19TH
Convento de São Francisco
Coimbra
Thursday, 17th Nov

10h00-12h00 – SPGH CLUB MEETINGS (concurrent)
  › CLUB 1: Cytogenetic and Molecular Genetics Club
     Chair: Eunice Matoso e Susana Ferreira
  › CLUB 2: Dysmorphology and Clinical Genetics Club
     Chair: Jorge Saraiva

14h00-14h15 – OPENING & WELCOME

14h15-15h00 – KEYNOTE LECTURE 1
  › DNA Methylation Episignatures for Diagnosis of Rare Disorders
     Beckim Sadikovic, Western University, London, Ontario, Canada

15h00-16h00 – Invited Symposium I (IS-I): GENETICS AND INBORN ERRORS OF METABOLISM

15h00-15h30 – ISI-1: Title to be announced. Clara Van Karnebeek, Department of Pediatrics, Radboud Centre for Mitochondrial Medicine, Radboud University Medical Centre, Nijmegen, Netherlands
15h30-16h00 – ISI-2: Title to be announced. Dulce Quelhas, Unidade de Bioquímica Genética, Centro De Genética Médica Jacinto De Magalhães, Centro Hospitalar Do Porto, Porto, Portugal

16h00-16h20 – Coffee-break

16h20-17h00 – POSTER VIEWING AND DISCUSSION

17h00-17h20 – CORPORATE SYMPOSIUM I. ILC

17h20-18h20 – 5 SELECTED ORAL PRESENTATIONS I (Basic Research)

18h20-18h40 – CORPORATE SYMPOSIUM II. HeartGenetics

18h45 – SPGH General Assembly
Friday 18th Nov

08h45-09h45 – 5 SELECTED ORAL PRESENTATIONS II (Clinical Research)

09h45-10h45 – BUILDING BRIDGES: SPGH and Associação de Diagnóstico Pré-natal

10h45-11h00 – Coffee-break

11h00-11h20 – CORPORATE SYMPOSIUM III. Unilabs
   WES VS TARGETED PANEL FOR DIAGNOSIS OF DEVELOPMENTAL DELAY: CLINICAL INSIGHTS FROM OUR EXPERIENCE. Rita Cerqueira, Laboratory Director, Unilabs Genetics

11h20-12h20 – Invited Symposium II (IS-II): CHROMATIN REMODELING AND GENE EXPRESSION IN HEALTH AND DISEASE.
   11h20-11h50 – ISII-1: Title to be announced. Pedro Prudêncio, Faculty of Medicine, University of Lisbon Lisboa, Portugal
   11h50-12h20 – ISII-2: Title to be announced. Margarida Gama Carvalho, Faculty of Sciences, University of Lisbon, Lisboa, Portugal

12h20-13h00 – KEYNOTE LECTURE 2
   Interpreting variation in the non-coding genome
   Nicky Whiffin, Wellcome Centre for Human Genetics, University of Oxford, Oxford, United Kingdom

13h05-14h00 – Lunch break

14h00-14h30 – POSTER VIEWING AND DISCUSSION

14h30-15h30 – Invited Symposium III. (IS-III) GENETIC AND PRECISION MEDICINE
   14h30-15h00 – ISIII-1: Title to be announced. Elfride de Baere, Center for Medical Genetics, Ghent University, Ghent, Belgium
   15h00-15h30 – ISIII-2: Title to be announced. Ana Teresa Maia, Faculty of Medicine and Biomedical Sciences, University of Algarve, Faro, Portugal
15h30-15h50 – CORPORATE SYMPOSIUM IV. Astrazeneca

15h50-16h10 – Coffee-break

16h10-17h20 – Panel Discussion (PDI): "POLIGENIC RISK SCORES"

16h10-16h20 – PDI -1: Clinical Genetics perspective
       Jorge Saraiva, Centro Hospitalar e Universitário de Coimbra – CHUC, Portugal

16h20-16h30 – PDI -2 Ophthalmology perspective
       Rufino Silva, Centro Hospitalar e Universitário de Coimbra – CHUC, Portugal

16h30-16h40 – PDI -3: Neurology perspective
       Miguel Pereira, Centro Hospitalar e Universitário de Coimbra – CHUC, Portugal

16h40-16h50 – PDI -4: Oncology perspective
       Manuel Teixeira, Portuguese Oncology Institute – Porto, Porto, Portugal

16h50-17h20 – Discussion

17h20-17h40 – CORPORATE SYMPOSIUM V. Janssen

20h00 – CONGRESS DINNER
Saturday 19th Nov

08h30-09h30 – 5-8 SELECTED ORAL PRESENTATIONS III (Clinical Cases)

09h30-10h30 – BIOETHICS DEBATE: “INTELIGÊNCIA ARTIFICIAL E SAÚDE – POTENCIALIDADES E LIMITAÇÕES ÉTICAS E TÉCNICAS NAS SUAS APLICAÇÕES, NOMEADAMENTE NA ÁREA DA GENÉTICA HUMANA”. Heloísa Santos, President of the Bioethics Commission of SPGH.

10h30-11h15 – Coffee-break and POSTER VIEWING

11h15-12h00 – KEYNOTE LECTURE 3
› Next generation-regeneration: iPSC as tools for lineage specification and disease modeling.
   Gustavo Mostoslavsky, Boston University School of Medicine, Boston, USA

12h00-12h15 – SPGH AWARD LECTURE

12h15-12h40 – SPGH AWARDS CEREMONY

12h40-13h00 – CLOSING SESSION