

Preliminary Programme



26TH ANNUAL MEETING

Sociedade Portuguesa
de Genética Humana

2022 NOV 17TH - 19TH
Convento de São Francisco
Coimbra



SPGH.NET



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: Personalized Medicine for Neurometabolic Diseases in 2022.** *Clara Van Karnebeek, Department of Pediatrics, Radboud Centre for Mitochondrial Medicine, Radboud University Medical Centre, Nijmegen, Netherlands*

15h30-16h00 – **ISI-2: Congenital Disorders of Glycosylation in Portugal – 20 years in a nutshell.** *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.** Key benefits of using Comprehensive Genomic Profiling (CGP) assays on tissue and liquid biopsy samples - A PGDx portfolio and cases studies review.
Patrick Hollender, Internacional Sales Director, PGDx

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.**

Fabiana Ramos, Medical Genetics Department, Paediatric Hospital of Coimbra, Centro Hospitalar e Universitário de Coimbra

Miguel Branco, President of the Portuguese Society of Prenatal Diagnosis

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-: RNA PROCESSING REVEALED BY NASCENT TRANSCRIPTOMICS.** *Pedro Prudêncio*, Faculty of Medicine, University of Lisbon Lisboa, Portugal

11h50-12h20 – **INFERRING MOLECULAR MECHANISMS FROM DISEASE MODEL TRANSCRIPTOMICS: LIMITATIONS AND PROMISES FROM NETWORK APPROACHES.** *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

13h05-13h25 – **CORPORATE SYMPOSIUM IV. Bionano Genomics With Lunch Box**

SEE MORE, KNOW MORE: HOW OGM PROVIDES ANSWERS

FOR RARE UNDIAGNOSED GENETIC DISEASES. *Alicia Bertolotti*, Bionano Genomics

13h25-14h00 – **Break Time**

14h00-14h30 – **POSTER VIEWING AND DISCUSSION**

14h30-15h30 – **Invited Symposium III (IS-III): GENETIC AND PRECISION MEDICINE**

14h30-15h00 – **MULTI-OMICS PROFILING, IN VITRO AND IN VIVO APPROACHES TO INTERPRET NON-CODING VARIATION IN RARE EYE DISEASES.** *Elfride de Baere*, Center for Medical Genetics, Ghent University, Ghent, Belgium





15h00-15h30 – **REGULATORY VARIANTS IN BREAST CANCER – FROM RISK TO PROGNOSIS.**

Ana Teresa Maia, Faculty of Medicine and Biomedical Sciences, University of Algarve, Faro, Portugal

15h30-15h50 – **CORPORATE SYMPOSIUM V.** 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 R. Teixeira, Portuguese Oncology Institute – Porto, Porto, Portugal

16h50-17h20 – **Discussion**

17h20-17h40 – **CORPORATE SYMPOSIUM VI.** Janssen

GENETIC TESTING IN PROSTATE CANCER – A NEW ERA

Moderador: *Pedro Madeira*, IPO Coimbra

- **Experiência de um Centro Português na testagem de Homologous Recombination Repair (HRR) no Cancro da Próstata**, *Manuel R. Teixeira*

20h00 – **CONGRESS DINNER**





Saturday 19th Nov

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

09h30-10h30 – **BIOETHICS DEBATE: ARTIFICIAL INTELLIGENCE AND NEW PREDICTIVE ANALYTICS – THE SCIENTIFIC AND ETHIC IMPACT IN PUBLIC HEALTH AND CLINICAL GENETICS. THE BENEFITS, THE CHALLENGES, THE WIDENING OF DISPARITY AND OTHER IMPORTANT RISKS.**

INTELIGÊNCIA ARTIFICIAL (IA) E A NOVA ANÁLISE PREDITIVA. IMPACTO ÉTICO E CIENTÍFICO EM SAÚDE PÚBLICA E GENÉTICA MÉDICA. BENEFÍCIOS, DESAFIOS E IDENTIFICAÇÃO DE RISCOS.

09h30-09h35 – **INTRODUCTION.** *Carolino Monteiro*, Faculty of Pharmacy of the University of Lisbon, Portugal

09h35-09h55 – **ARTIFICIAL INTELLIGENCE IN HEALTH CARE: SOME ETHICAL AND SOCIAL ASPECTS.**

Mário Figueiredo, Instituto Superior Técnico, University of Lisbon, Portugal

09h55-10h05 – **ARTIFICIAL INTELLIGENCE IN GENOMICS.** *José Carlos Ferreira*,

Centro Hospitalar Universitário Lisboa Norte

10h05-10h10 – **ETHICAL REFLECTIONS AND SOME QUESTIONS.** *Célia Ventura*, Department of Human Genetics, National Institute of Health Doutor Ricardo Jorge, Portugal

10h10-10h30 – **GENERAL DISCUSSION.** *Heloísa Santos*, Bioethics Committee President

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**

