



# 26<sup>TH</sup> ANNUAL MEETING

—  
Sociedade Portuguesa  
de Genética Humana  
—

2022 NOV 17<sup>TH</sup> - 19<sup>TH</sup>  
Convento de São Francisco  
Coimbra



SPGH.NET



## Scientific Meeting Program / Programa Científico

### Thursday, 17<sup>th</sup> Nov

10h00-12h00 – **SPGH CLUB MEETINGS (concurrent)**

› **CLUB 1: Cytogenetic and Molecular Genetics Club**

Anfiteatro do Hospital Pediátrico

Ed. do Hospital Pediátrico de Coimbra, Piso 0

**Chairs:** *Eunice Matoso and Susana Ferreira*

› **CLUB 2: Dymorphology and Clinical Genetics Club**

Sala da ACREDITAR – Edifício no Hospital Pediátrico de Coimbra

**Chair:** *Jorge Saraiva*

14h00-14h15 – **OPENING & WELCOME**

*Lina Ramos, Ilda Patrícia Ribeiro, Rosário Pinto Leite, Joana Barbosa de Melo, Presidente da Câmara Municipal de Coimbra, Conselho de Administração do CHUC, Direção da FMUC*

14h15-15h00 – **KEYNOTE LECTURE 1**

› **DNA Methylation Episignatures for Diagnosis of Rare Disorders**

*Bekim Sadikovic, Western University, London, Ontario, Canada*

**Chairs:** *Lina Ramos and Sérgio Sousa*

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

**Chairs:** *Rosário Pinto Leite and Paula Jorge*

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)**

**Chairs:** *Clévio Nobrega and Ilda Patrícia Ribeiro*

	<b>Title</b>	<b>Author</b>
OPI-1 17h20-17h30	Disruption of regulatory elements in the CDH1 vicinity as a potential HDGC cause, increased penetrance and early onset cancer	<i>Celina São José</i>
OPI-2 17h30-17h40	DDIT4 and TRIM13 Transcript Levels are Blood-Based Biomarkers of Early Stages of Machado-Joseph Disease/ Spinocerebellar Ataxia Type 3	<i>Ana F. Ferreira</i>
OPI-3 17h40-17h50	DAB1-antisense Expression at the Spinocerebellar Ataxia Type 37 (SCA37) Locus and Disease	<i>Catarina C. Rodrigues</i>
OPI-4 17h50-18h00	Modelling Machado-Joseph disease in patient-derived isogenic lines by CRISPR/CAS9-MEDIATED correction of ATXN3-CAG expansion	<i>Pedro Perdigão</i>
OPI-5 18h00-18h10	tRNA epitranscriptome reprogramming in Alzheimer’s disease correlates with protein aggregation burden	<i>Ana R. Soares</i>

**18h20-18h40 – Corporate symposium II: HeartGenetics.**

**PHARMACOGENETICS: 7 DECADES LATER.** *Daniel Luís, HeartGenetics - Portugal*

**18h45 – SPGH General Assembly**

**Chairs:** *Lina Ramos, Ilda Patrícia Ribeiro and Rosário Pinto Leite*

**Friday 18<sup>th</sup> Nov**

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

**Chairs:** *Eunice Matoso and Sofia Dória*

	<b>Title</b>	<b>Author</b>
OPII-1 08h45-08h55	Impact of genomic medicine for critical ill infants: a retrospective study of a reference center in Lisbon, Portugal	<i>Susana Lemos Ferreira</i>
OPII-2 08h55-09h05	Expanding The Genetics of Hearing Loss: The Largest Portuguese Cohort and The First in Pediatric Age	<i>Sofia Nunes</i>
OPII-3 09h05-09h15	The contribution of PRS for small vessel disease and age-related DNA methylation and gene expression changes to cognitive aging	<i>Sonya Neto</i>
OPII-4 09h15-09h25	Whole-Genome Sequencing analysis to identify candidate HDGC genetic modifiers for the CDH1 c.1901 variant	<i>João Fonseca</i>
OPII-5 09h25-09h35	CTNNA1 germline variants: disease spectrum extending beyond Hereditary Diffuse Gastric Cancer	<i>Silvana Lobo</i>



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

**Chair:** *Lina Ramos*

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.**

**Chairs:** *Sebastião Rodrigues and Peter Jordan*

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

**Chairs:** *Gabriela Soares and Joaquim de Sá*

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**

**Chairs:** *Isabel Marques Carreira and Jorge Saraiva*

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**

**DIFFERENT PERSPECTIVES IN GENETIC LITERACY IN ONCOLOGY.** *Carla Oliveira (IPATIMUP)*

*Mariana Soeiro e Sá (CHULN); Enfermeira Tamara Milagres (EVITA)*

15h50-16h10 – **Coffee-break**

16h10-17h20 – **Panel Discussion (PDI): “POLIGENIC RISK SCORES”**

**Chairs:** *Lina Ramos and Joana Barbosa de Melo*

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 Tábuas-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**

**Chair:** *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 19<sup>th</sup> Nov

08h30-09h35 – **9 SELECTED ORAL PRESENTATIONS III (Clinical Cases)**

**Chairs:** *Joana Salgado and Patrícia Dias*

	<b>Title</b>	<b>Author</b>
OPIII-1 08h30-08h37	BIALLELIC DYNC2H1 SPLICING VARIANTS CAUSING PRENATAL SHORT-RIB POLYDACTYLY SYNDROME: CLINICAL, RADIOLOGICAL, AND HISTOPATHOLOGIC FEATURES	<i>Rita Quental</i>
OPIII-2 08h37-08h44	MOHR-TRANEBJAERG SYNDROME: HEARING LOSS AS THE ONSET OF AN INSIDIOUS DISORDER WITH HIGH RECURRENCE RISK	<i>Maria Abreu</i>
OPIII-3 08h44-08h51	NF2-RELATED SCHWANNOMATOSIS – AN ATYPICAL CASE PRESENTATION	<i>Marta P. Soares</i>
OPIII-4 08h51-08h58	ERF HAPLOINSUFICIENCY: EXPLORING VARIABLE EXPRESSIVITY, INCOMPLETE PENETRANCE, AND OVERLAPPING DIAGNOSIS	<i>Mafalda Melo</i>
OPIII-5 08h58-09h05	TWO CASE REPORT OF A NEW RECOGNIZABLE SYNDROME - DEGCAGS (DEVELOPMENTAL DELAY WITH GASTROINTESTINAL, CARDIOVASCULAR, GENITOURINARY, AND SKELETAL ABNORMALITIES)	<i>Mafalda Santos</i>
OPIII-6 09h05-09h12	DESCRIPTION OF TWO NEW CASES OF WIEACKER-WOLFF SYNDROME (ZARD) ASSOCIATED TO DE NOVO ZC4H2 SPLICING VARIANTS	<i>Diogo Fernandes da Rocha</i>
OPIII-7 09h12-09h19	46,XY DISORDER OF SEX DEVELOPMENT DUE TO LHCGR COPY LOSS IN AOH REGION	<i>Sónia Custódio</i>
OPIII-8 09h19-09h26	TREACHER COLLINS SYNDROME ASSOCIATED WITH POLR1D – A Milder PHENOTYPE? REPORT OF TWO FAMILIES	<i>Mariana Tomásio Neves</i>
OPIII-9 09h26-09h33	TAF2 RELATED TO THE TRANSCRIPTION FACTOR TFIID: A NEW FAMILY AND REVIEW OF THE LITERATURE	<i>Catarina S. Rosas</i>

09h35-10h35 – **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.

**Chair:** *Heloísa Santos*

09h35-09h40 – **INTRODUCTION.** *Carolino Monteiro*, Faculty of Pharmacy of the University of Lisbon, Portugal

09h40-10h00 – **ARTIFICIAL INTELLIGENCE IN HEALTH CARE: SOME ETHICAL AND SOCIAL ASPECTS.**

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

10h00-10h10 – **ARTIFICIAL INTELLIGENCE IN GENOMICS.** *José Carlos Ferreira*, Centro Hospitalar Universitário Lisboa Norte

10h10-10h15 – **ETHICAL REFLECTIONS AND SOME QUESTIONS.** *Célia Ventura*, Department of Human Genetics, National Institute of Health Doutor Ricardo Jorge, Portugal

10h15-10h35 – **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*

**Chairs:** *Carla Oliveira and Joana Barbosa de Melo*

12h00-12h15 – **SPGH AWARD LECTURE**

**Chairs:** *Lina Ramos and Joana Barbosa de Melo*

12h15-12h40 – **SPGH AWARDS CEREMONY**

**Chairs:** *Lina Ramos and Joana Barbosa de Melo*

12h40-13h00 – **CLOSING SESSION**

**Chairs:** *Lina Ramos, Ilda Patrícia Ribeiro and Rosário Pinto Leite*