ANNUAL MEETING

Virtual

SOCIEDADE PORTUGUESA DE GENÉTICA HUMANA

18 - 19
NOV 2021

PRELIMINARY PROGRAMME
09:00  OPENING & WELCOME

09:15  25TH ANNIVERSARY SESSION
      Chairs: Jorge Pinto Basto, Carla Oliveira
      AMÂNDIO S. TAVARES AND THE FOUNDATIONS OF HUMAN GENETICS IN PORTO
      Jorge Sequeiros (i3S, Porto, PT)
      THE BEGINNINGS OF MEDICAL GENETICS AND THE CREATION OF SPGH - CONTRIBUTION TO THE HISTORY OF ITS FOUNDATION
      Heloísa Santos, (1st President of the SPGH, PT)
      KEYNOTE: DNA METHYLATION LANDSCAPES OF BREAST CANCER REVEAL EPIGENETIC INSTABILITY AND CIS REGULATION
      Carlos Caldas, (CRUK Cambridge Institute, Cambridge, UK)

10:15  COFFEE-BREAK / ONLINE EXHIBITION

10:25  INVITED SYMPOSIUM I (IS-I):
      CHROMATIN REMODELLING IN HEALTH AND DISEASE
      Chairs: Sérgio Sousa, Sebastião Rodrigues
      ISI-1: THE PANCREAS REGULOME: FROM CAUSALITY TO PREDICTION OF NON-CODING MUTATIONS IN HUMAN PANCREATIC DISEASES
      José Bessa (i3S, Porto, PT)

      ISI-2: ROLE OF BAF SWI/SNF CHROMATIN REMODELLERS IN NEURODEVELOPMENT AND ID
      Cristina Dias (CRICK, London, UK)
11:25  CORPORATE SYMPOSIUM
Prenatal Trio Exome Sequencing for Fetal Ultrasound Anomalies
Detlef Trost

11:45  CORPORATE SYMPOSIUM
Gene Therapy – From Genesis to Treatment

12:25  CORPORATE SYMPOSIUM
Genetic Diagnosis of Gastrointestinal Hereditary Cancer Syndromes: NGS Update
Cristina Albuquerque

12:45  LUNCH BREAK

13:10  POSTER VIEWING AND DISCUSSION / ONLINE EXHIBITION

14:40  SELECTED ORAL PRESENTATIONS I (5X BASIC RESEARCH)
Chairs: Joana Melo, Paula Jorge

15:55  PANEL DISCUSSION (PDI):
Emergent Challenges in Human Genetics
Chairs: Ana Berta Sousa, Jorge Saraiva

PDI-1: Carrier Screening in Portugal: Towards a Scientifically Sound, Ethically Rigorous, Socially Responsible Programme
Jorge Sequeiros (i3S, Porto, PT)

PDI-2: Technical Standards of the Interpretation and Reporting of Constitutional Copy-Number Variants
Erin Rooney Riggs (USA)

16:55  COFFEE-BREAK / POSTER VIEWING AND DISCUSSION / ONLINE EXHIBITION

17:10  SELECTED ORAL PRESENTATIONS II (5X CLINICAL RESEARCH)
Chairs: Sofia Dória, Gabriel Miltényi-Miltenberger

18:25  GENERAL ASSEMBLY
08:30  SELECTED ORAL PRESENTATIONS III (5 X CLINICAL CASE REPORTS)
Chairs: Gabriela Soares, Lina Ramos

09:30  BIOETHICS DEBATE (BE): ETHICAL IMPERATIVE – THE IMPORTANCE OF QUALITY ON DIAGNOSTICS AND PRACTICE IN GENETIC MEDICINE
Chairs: Célia Ventura, Carolino Monteiro

BE-1:  THE CLINICAL GENETICIST PERSPECTIVE
Lina Ramos (CHUC, Coimbra, PT)

BE-2:  THE LABORATORY GENETICIST PERSPECTIVE
Belinda Xavier (CHUV, Lausanne, CH)

BE-3:  THE GENETICS SCIENTIST PERSPECTIVE
Carla Oliveira (i3S/Ipamup, Porto, PT)

GENERAL DISCUSSION
Heloísa Santos, 1st President of the SPGH (PT)

10:30  COFFEE-BREAK / ONLINE EXHIBITION

10:45  CORPORATE SYMPOSIUM
ACHONDROPLASIA TREATMENT

11:05  CORPORATE SYMPOSIUM
ADVANCES IN WGS APPLICATIONS: LATEST COLLABORATIONS AND INNOVATIONS
Martin Boura

11:45  CORPORATE SYMPOSIUM
DISTROFIAS DA RETINA: A ATUALIDADE DO DIAGNÓSTICO E TRATAMENTO, E PERSPETIVAS FUTURAS”
Ana Luisa Carvalho
12:05  LUNCH BREAK

12:30  POSTER VIEWING AND DISCUSSION / ONLINE EXHIBITION

14:00  INVITED SYMPOSIUM II (ISII):
STUDY MODELS AND TREATMENTS IN GENETIC DISEASES
*Chairs: Carla Oliveira, Rosário Santos*

**ISII-1:** SPINAL MUSCULAR ATROPHY TREATMENT UPDATES
*Eduardo Tizzano (VHIO, Barcelona, ES)*

**ISII-2:** NEUROFIBROMATOSIS TREATMENT UPDATES
*João Passos (IPOL, Lisboa, PT)*

**ISII-2:** SYNTHETIC LETHALITY: PROMISES AND HURDLES IN CANCER TREATMENT
*Chris Lord (CRUK, London, UK)*

15:25  KEYNOTE LECTURE
*Chair: João Gonçalves*

TELOMERE MAINTENANCE, CHROMOSOMAL INTEGRITY AND THE SPECTRUM OF HUMAN DISEASES”
*Luis Batista (Siteman Cancer Center, Washington, USA)*

16:00  COFFEE-BREAK / POSTER VIEWING AND DISCUSSION / ONLINE EXHIBITION

16:20  SPGH AWARD LECTURE

16:50  SPGH AWARDS CEREMONY

17:15  CLOSING SESSION