25 ANOS

Virtual ANNUAL MEETING

SOCIEDADE PORTUGUESA DE GENÉTICA HUMANA

18 - 19
NOV 2021

PRELIMINARY PROGRAMME

SPGH
www.spgh.net

09:00 **OPENING & WELCOME**

09:15 **25TH ANNIVERSARY SESSION**

Chair: Heloísa Santos

KEYNOTE: FINDING GENOMIC AND EPIGENOMIC SIGNATURES THAT DRIVE BREAST CANCER AND ITS RESPONSE TO TREATMENT

Carlos Caldas (CRUK Cambridge Institute, Cambridge, UK)

HISTÓRIA DA GENÉTICA MÉDICA EM PORTUGAL

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

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 SESSION**
PRENATAL TRIO EXOME SEQUENCING FOR FETAL ULTRASOUND ANOMALIES
Detlef Trost

grupo I.L.C.

11:45 **CORPORATE SESSION**
ADVANCES IN WGS APPLICATIONS: LATEST COLLABORATIONS AND INNOVATIONS
Martin Boura

Quilaban
uma fórmula de vida

12:15 **CORPORATE SESSION**
GENETIC DIAGNOSIS OF GASTROINTESTINAL HEREDITARY CANCER SYNDROMES: NGS UPDATE
Cristina Albuquerque

SRO SOQUÍMICA

12:35 LUNCH BREAK

13:00 **POSTER VIEWING AND DISCUSSION / ONLINE EXHIBITION**

14:30 **SELECTED ORAL PRESENTATIONS I** (5X BASIC RESEARCH)
Chairs: Joana Melo, Paula Jorge

15:45 **PANEL DISCUSSION (PDI):**
EMERGENT CHALLENGES IN HUMAN GENETICS
Chairs: Ana Berta Sousa, Jorge Saraiva

PDI-1: GENETIC CARRIER SCREENING IN PORTUGAL
Jorge Sequeiros (i3S, Porto, PT)

PDI-2: TECHNICAL STANDARDS OF THE INTERPRETATION AND REPORTING OF CONSTITUTIONAL COPY-NUMBER VARIANTS
Erin Rooney Riggs (USA)

16:45 COFFEE-BREAK / POSTER VIEWING AND DISCUSSION / ONLINE EXHIBITION

17:00 **SELECTED ORAL PRESENTATIONS II** (5X CLINICAL RESEARCH)
Chairs: Sofia Dória, Gabriel Miltényi-Miltenberger

18:15 **GENERAL ASSEMBLY**

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

09:30 **BIOETHICS DEBATE (BE): QUALIDADE COMO IMPERATIVO ÉTICO**
Chair: Heloísa Santos

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/Ipatimup, Porto, PT)

GENERAL DISCUSSION

10:30 COFFEE-BREAK / ONLINE EXHIBITION

10:45 **CORPORATE SYMPOSIUM**
ACHONDROPLASIA TREATMENT

11:05 **CORPORATE SYMPOSIUM**

11:45 **CORPORATE SYMPOSIUM**
DISTROFIAS DA RETINA: A ATUALIDADE DO DIAGNÓSTICO E TRATAMENTO, E PERSPETIVAS FUTURAS"
Ana Luísa 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**

SPGH

Sociedade Portuguesa
de Genética Humana

SPGH CONTACTS
Faculdade de Medicina - Polo I
Universidade de Coimbra
Rua Larga, 3004-504, COIMBRA
spgh1997@gmail.com
www.spgh.net

SECRETARIAT CONTACTS
VERANATURA
Rua Augusto Macedo, 12-D Esc. 2 / 3,
1600-794 LISBOA - PORTUGAL
Tels.: +351 217 120 778 / 79
spgh@veranatura.pt

www.spgh.net