

28TH ANNUAL MEETING

SOCIEDADE PORTUGUESA
DE GENÉTICA HUMANA

5 - 7
DECEMBER
2024



INSTITUTO SUPERIOR DE ENGENHARIA

ISEP PORTO

Rua António Bernardino Almeida, 431
4200-529 Porto



Preliminary Programme



Thursday 5th Dec

09h00-14h00 › Registration

10h00-12h00 › PRE-MEETING CONCURRENT SESSIONS

- › EDUCATIONAL SESSION 1: Scientific Writing
- › CLUB 1: Cytogenetics and Molecular Genetics – Case discussion
- › EDUCATIONAL SESSION 2: Classification of variants – Rules and criteria
- › CLUB 2: Dysmorphology and Clinical Genetics

12h30-13h30 › CORPORATE MEETINGS (concurrent in 4 or 5 rooms)

13h30-14h00 › **Lunch break** – Lunch offered to attendees of corporate meetings

14h05-14h15 › OPENING AND WELCOME

14h15-14h50 › KEYNOTE LECTURE 1

Chair: TBA

“Single-cell, whole-embryo phenotyping of mammalian developmental disorders”

Malte Spielmann (Max Planck Institute for Human Genetics, Germany)

14h15-14h50 › INVITED SYMPOSIUM I (IS-I): *Genetics of Neurological Disorders*

Chair: TBA

14h50-15h20 › INVITED TALK ISI-1: *“Genetics and emerging treatments for neurometabolic diseases”*

Fanny Mochel (Sorbonne Université - Hôpital de la Pitié-Salpêtrière, Paris, France)

15h20-15h50 › INVITED TALK ISI-2: *“Trinity of neuroscience, genetics, and bioinformatics”*

Friederike Ehrhart (Maastricht University, The Netherlands)

15h50-16h10 › CORPORATE SYMPOSIUM (CSI): RESERVED

16h10-16h30 › Coffee-break

16h30-17h30 › POSTER VIEWING AND DISCUSSION, Session 1

17h10-18h20 › SELECTED ORAL PRESENTATIONS I: Basic Research

Chair: TBA

- › 17h30-17h40 › Selected abstract 1
- › 17h40-17h50 › Selected abstract 2
- › 17h50-18h00 › Selected abstract 3
- › 18h00-18h10 › Selected abstract 4
- › 18h10-18h20 › Selected abstract 5

18h30 › SPGH GENERAL ASSEMBLY



Friday 6th Dec

09h00-10h00 › SELECTED ORAL PRESENTATIONS II: Clinical Research

Chair: TBA

- › 09h00-09h10 › Selected abstract 1
- › 09h10-09h20 › Selected abstract 2
- › 09h20-09h30 › Selected abstract 3
- › 09h30-09h40 › Selected abstract 4
- › 09h40-09h50 › Selected abstract 5

10h00-11h00 › BUILDING BRIDGES: *Hereditary Cancer Network in Portugal*

Chair: TBA

Program in preparation

11h00-12h00 › Coffee-break / Poster Viewing and Discussion, Session 2

12h00-13h00 › INVITED SYMPOSIUM II (ISII): Update on preimplantation genetic testing and prenatal diagnosis in Portugal

Chair: TBA

12h00-12h30 › INVITED TALK ISII-1: "Preimplantation genetic testing in Portugal"

› *Filipa Carvalho (Serviço de Genética, Department of Pathology, Faculty of Medicine, University of Porto, Porto, Portugal)*

12h30-13h00 › INVITED TALK ISII-2: "Prenatal diagnosis in Portugal"

Inês Carvalho (Unidade Local de Saúde São José, Lisbon, Portugal)

13h00-14h00 › Lunch break

14h00-15h00 › POSTER VIEWING AND DISCUSSION, Session 3

15h00-16h00 › INVITED SYMPOSIUM III (ISIII): Cancer Genetics

Chair: TBA

15h00-15h30 › Invited talk ISIII-1: "Genetics of Li-Fraumeni"

Gaëlle Bougeard (Université de Rouen, France)

15h30-16h00 › Invited talk ISIII-2: "The urothelial gene regulatory network: understanding biology to improve bladder cancer management"

Francisco X. Real (CNIO, Madrid, Spain)

16h00-16h30 › Coffee-break

16h30-16h50 › SELECTED ORAL PRESENTATIONS III: Clinical Cohorts

Chair: TBA

16h30-16h40 › Selected abstract 1

16h40-16h50 › Selected abstract 2

16h30-16h50 › ROUND TABLE: Bridging imaging and genetics

Chair: TBA

João Soares Fernandes (Hospital de Braga, Braga, Portugal)

José Almeida (Champalimaud Foundation, Lisbon, Portugal)

17h45-18h45 › INVITED SYMPOSIUM IV (ISIV): Understanding and treating disease

Chair: TBA

17h45-18h15 › Invited talk ISIV-1: "Genotype-phenotype correlations in Parkinson Disease"

Joana Damásio (Universidade do Porto Instituto de Ciências Biomédicas Abel Salazar, Portugal)

17h45-18h45 › Invited talk ISIV-2: "Ataxia with giant axonopathy in Acbd5-deficient mice halted by adeno-associated virus gene therapy"

Pedro Brites (I3S, Universidade do Porto, Portugal)

20h00

GALA DINNER



Saturday 7th Dec

08h40-09h30 › SELECTED ORAL PRESENTATIONS III: Clinical cases

Chair: TBA

08h40-08h50 › Selected abstract 1

08h50-09h00 › Selected abstract 2

09h00-09h10 › Selected abstract 3

09h10-09h20 › Selected abstract 4

09h20-09h30 › Selected abstract 5

09h30-10h30 › BIOETHICS DEBATE: "Lack of access to healthcare in rare diseases"

Chair: *Lina Ramos*

Program under preparation

10h30-10h50 › CORPORATE SYMPOSIUM II (CSII) -

10h50-11h25 › Coffee-break

11h25-12h00 › KEYNOTE LECTURE 2

Chair: TBA

"Genoprotective interventions for healthspan extension"

Elsa Logarinho (i3S, Porto, Portugal)

12h00-12h25 › SPGH Award Lecture

12h25-12h45 › SPGH Awards Ceremony

12h45-13h00 › Closing Session