



28ª Reunião Anual / 28th Annual Meeting

5 - 7 December 2024

Preliminary Programme

Thursday 5th Dec

09h00-14h00 Registration

10h00-12h00 Pre-meeting Concurrent Sessions

- EDUCATIONAL SESSION 1: *Scientific Writing*
- CLUB 1: *Cytogenetics and Molecular Cytogenetics – 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)*

14h50-15h50 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*

17h00-17h40 **ROUND TABLE: Bridging imaging and genetics**

Chair: TBA

➤ **Ana Filipa Gerardo** (CHVNG/E, Porto, 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)

18h15-18h45 *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

Pogram 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