



## 28ª Reunião Anual / 28<sup>th</sup> Annual Meeting

5 - 7 December 2024

### Preliminary Programme

#### Thursday 5<sup>th</sup> 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 IS-1: "Genotype-phenotype correlations in Parkinson Disease"*

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

15h20-15h50 *Invited talk IS-2: "Ataxia with giant axonopathy in Acbd5-deficient mice halted by adeno-associated virus gene therapy"*

- **Pedro Brites** (I3S, Universidade do Porto, Portugal)

**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 6<sup>th</sup> 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)
- **Regina Beets-Tan** (The Netherlands Cancer Institute, Amsterdam, Netherlands) - *To be confirmed*

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

Chair: TBA

17h45-18h15 *Invited talk ISIV-1: "Genetics and emerging treatments for neurometabolic diseases"*

- **Fanny Mochel** (Sorbonne Université - Hôpital de la Pitié-Salpêtrière, Paris, France) - *To be confirmed*

18h15-18h45 *Invited talk ISIV-2: "Trinity of neuroscience, genetics, and bioinformatics"*

- **Friederike Ehrhart** (Maastricht University, The Netherlands)

**20h00 GALA DINNER**

## Saturday 7<sup>th</sup> 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