# 30ª Reunião Anual /30th *Annual Meeting*

## 19 - 21 November 2026

## Preliminary Programme

**Thursday 19th Nov**

**09h00** **Registration**

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| **Main auditorium** | **PRE-MEETING SESSION** |
| **9h30-10h20** | *Deep generative models of large-scale functional genetic screens for drug target discovery*<br>➢ **Emanuel Gonçalves** – Instituto Superior Técnico, Portugal |
| **10h20-11h10** | *Using genetics and genomics at scale to identify candidate drug targets and deliver precision medicine for inflammatory bowel disease*<br>➢ **Carl Anderson** - Welcome Sanger Institute, UK |
| **11h10- 12h00** | *TBA*<br>➢ **Manolis Kellis** - Massachusetts Institute of Technology (MIT), USA |
| **12:00-12h30** | *Final Discussion*<br>Chair: Carla Oliveira, i3S and IPATIMUP - University of Porto |
| **Room 1** | **CYTOGENETICS AND MOLECULAR GENETICS CLUB (session in PT)** |
| **10h30 – 11h00** | *Diagnóstico Molecular das Síndromes de Imprinting: Desafios, Estratégias e Casos Complexos.*<br>➢ Tba - INSA, Departamento de Genética Humana, Laboratório de Citogenética<br>Moderação: Hildeberto Correia, INSA |
| **11h00 – 11h30** | *Problemáticas na classificação da patogenicidade de variantes segundo diferentes guidelines/recomendações.*<br>➢ **Pedro Rodrigues** – INSA, Departamento de Genética Humana, Laboratório de Genética Molecular<br>Moderação: João Gonçalves, INSA |
| **11h30 – 12h30** | *Quando a IA falha: o impacto da validação especializada na genómica clínica.*<br>➢ **Lara Neto** - Unidade Local de Saúde de Santa Maria, Laboratório de Genética<br>➢ Tba - Centro de Genética Laboratorial Germano de Sousa |

Moderação: Ana Pereira, Centro de Medicina Laboratorial Germano de Sousa / Ana Sousa, Unidade Local de Saúde de Santa Maria

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| **Room 2** | **DYSMORPHOLOGY AND CLINICAL GENETICS CLUB (session in PT)** |
| **10h:30 – 12h30** | *BAFopatias e outras doenças da cromatina.*<br>➢ **Cristina Dias -** King's College London e Guy's and St Thomas NHS Foundation Trust<br>Moderação: Ana Berta Sousa, Unidade Local de Saúde de Santa Maria / Margarida Venâncio, Unidade Local de Saúde de São José |
| **12h40-13h00** | **CORPORATE SYMPOSIUM** |
| **13h00-14h00** | **Lunch break –** *Lunch offered to attendees of corporate meeting* |
| **14H05-14h15** | **OPENING AND WELCOME** |
| **14h15-14h50** | **KEYNOTE LECTURE 1 -Title tba**<br>➢ **Manolis Kellis** - Massachusetts Institute of Technology (MIT), USA |
| **14h50-15h50** | **Invited Symposium I (IS-I): Mitochondrial diseases** |
| 14h50-15h20 | *Invited talk ISI-1: The diagnostic journey in mitochondrial disease: from clinical suspicion to metabolomics*<br>➢ **Margarida Coelho -** Centro Hospitalar do Porto |
| 15h20-15h50 | *Invited talk ISI-2: Integrative Omics Analyses to Improve Diagnostic Rates of Mendelian Disorders*<br>➢ **Holger Prokisch** - Institute of Human Genetics, Technical University Munich |
| **15h50-16h10** | **CORPORATE SYMPOSIUM – UNILABS**<br>*Integrating Artificial Intelligence into NGS variant reporting: CGC Genetics Experience from 30k exomes* |
| **16h10-17h30** | **Coffee-break** |
| **17h30-18h20** | **SELECTED ORAL PRESENTATIONS I:** Clinical research |
| **18h30** | **SPGH GENERAL ASSEMBLY** |

**Friday 20th Nov**

**09h00-10h00** **SELECTED ORAL PRESENTATIONS II:** Basic Research

**10h00-11h00** ***PANEL:*** *A genética humana na jornada da pessoa com doença rara* (session in PT)

*TBC*

**11h00-12h00** **Coffee-break / Poster Viewing and Discussion**

**12h00-13h00** **INVITED SYMPOSIUM II (ISII): Therapeutic Innovation in Rare Genetic Diseases**

**12h00-12h30** *Invited talk ISII-1: Gene therapy for DFNB9 hearing loss restores the function of otoferlin, a component of the auditory ribbon synapse*

- **Ignacio del Castillo -** Servicio de Genética, Hospital Universitario Ramón y Cajal, IRYCIS, Madrid, Spain

**12h30-13h00** *Invited talk ISII-2: The Changing Landscape of Cystic Fibrosis: Emerging CFTR Biology and Precision Theranostics*

- **Margarida Amaral -** BioISI – Biosystems & Integrative Sciences Institute, Faculty of Sciences, University of Lisboa, Lisboa Portugal

**13h00-14h30** **Lunch break & Poster Viewing**

**14h30-15h05** **KEYNOTE LECTURE 2**

*Using genetics and genomics at scale to identify candidate drug targets and deliver precision medicine for inflammatory bowel disease*

- **Carl Anderson** - Wellcome Sanger Institute, Wellcome Genome Campus

**15h05-16h05** **BIOETHICS DEBATE:** Title tba

**16h05-16h30** **Coffee-break**

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

**16h50-17h50** **INVITED SYMPOSIUM III (ISIII): Cancer predisposition throughout life**

**16h50-17h20** *Invited talk ISIV-1: Improving identification of genetic cancer predisposition in children with cancer: the advances and pitfalls of tumor-normal next generation sequencing*

- **Marjolijn Jongmans** - Radboud University Medical Center, Netherlands

**17h20-17h50** *Invited talk ISIV-2: The HDGC Puzzle: Genetics, Expressivity, Lifetime Risk, and Clinical Practice*

- **Carla Oliveira** - i3S and IPATIMUP, University of Porto

**17h50-18h00** **Anúncio das celebrações de 7 de dezembro (session in PT)**

- **Heloísa Santos** – Coordenadora das celebrações

**18h00-18:30** **Sessão de homenagem aos novos sócios honorários e beneméritos (session in PT)**

**20h00** **GALA DINNER**

## Saturday 21th Nov

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

**09h30-10h30** **BUILDING BRIDGES:** Centro de Referência da Paramiloidose

**TBA**

- **Dra. Isabel Conceição** - Centro de Referência da Paramiloidose

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

**10h50-11h25** **Coffee-break**

**11h25-12h00** **KEYNOTE LECTURE 3**

*Addressing dystrophin deficiency in muscle and brain in Duchenne muscular dystrophy*

- **Francesco Muntoni** - University College London Great Ormond Street Institute of Child Health and Queen Square Institute of Neurology, London UK

**12h00-12h25** **SPGH Award Lecture**

**12h25-12h45** **SPGH Awards Ceremony**

**12h45-13h00** **Closing Session**

## Coffee Break Sessions

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| Friday 20th, 11:00-11:30 | Room 1 | Roche<br>*TBA* |
| Friday 20th, 11:30-12:00 | Room 1 | Diagnostica Longwood<br>*Advances and Cutting-Edge Workflows in Clinical Genomics and Oncology* |
| Friday 20th, 16:05-16:30 | Room 1 | Varvis<br>*TBA* |