29.ª REUNIÃO ANUAL
29th ANNUAL MEETING

20-22 NOVEMBER 2025

CONVENTO SÃO FRANCISCO - COIMBRA

Preliminary Program

ORGANIZAÇÃO

SPGH
Sociedade Portuguesa de Genética Humana

THURSDAY 20th NOVEMBER

09h00-14h00 ▸ **REGISTRATION**

10h00-12h00 ▸ **PRE-MEETING PARALLEL SESSIONS**

- **PROFESSIONAL** (in Portuguese)
- **Session 1:** *Cytogenetics and Molecular Genetics Club*
  Chairs: Isabel Carreira | Teresa Fidalgo
- **Session 2:** *Dysmorphology and Clinical Genetics Club*
  Chairs: Joana Rosmaninho-Salgado, ULS Coimbra | Sadaf Farooqi, University of Cambridge
- **EDUCATIONAL**
- **Workshop 1:**
  Genetic testing interpretation and report – beyond ACMG classification
  Júlia Baptista, [1]Molecular Pathology Department, King's College Hospital, Synnovis, London; [2]Faculty of Life Sciences and Medicine, King's College London, UK
- **Workshop 2:**
  Gene therapy – from basic to advanced
  Luís Pereira de Almeida, Rui Nobre, CNC-UC & CIBB & Centro de Excelência em Terapia Genética (GeneT), Coimbra, Portugal

12h10-14h00 ▸ **PRE-CONGRESS PLENARY SESSION**

12h10-12h30 ▸ **CORPORATE SYMPOSIUM I ILC**
"Redefining Sequencing: From Genomics to 5D Multiomics with AVITI24"
Antonio Fadda, Field Applications Scientist, Element Biosciences

12h30-13h00 ▸ *Cooperation Protocol Signature Session (in Portuguese)*
Sérgio B. Sousa, President SPGH | Peter Jordan & Milena Paneque, CPPEG-SPGH | Mónica Correia, Sociedade Portuguesa de Literacia para a Saúde (SPLS) | Paulo Gonçalves, União das Associações das Doenças Raras de Portugal (RD-Portugal) | Rui Barros Silva, Federação das Doenças Raras de Portugal (FEDRA)

13h00-14h00 ▸ **LUNCH WITH PATIENT ASSOCIATIONS**

14h00-14h15 ▸ **OPENING AND WELCOME**
Sérgio B. Sousa, President SPGH 2025 | **Paula Jorge**, Chair Scientific Committee

14h15-14h50 ▸ **KEYNOTE LECTURE 1**
Chairs: Carla Oliveira | Paula Jorge
"Genome editing methods to interrogate variant function at scale"
Gregory Findlay, The Genome Function Laboratory, The Francis Crick Institute, London, United Kingdom

14h50-15h50 ▸ **INVITED SYMPOSIUM I:** Gene therapy for eye disorders
Chairs: Ana Luísa Carvalho | Rui Nobre
14h50-15h20 ▸ *Invited talk ISI-1: "Gene Therapy for Inherited Retinal Diseases"*
Bart Leroy, Department of Ophthalmology, Ghent University Hospital, Ghent, Belgium
15h20-15h50 ▸ *Invited talk ISI-2: "Gene Therapy in Portugal: Luxturna Outcomes and the Role of IRD-PT"*
João Pedro Marques, Department of Ophthalmology, ULS Coimbra, Coimbra, Portugal

15h50-16h10 ▸ **CORPORATE SYMPOSIUM II QUILABAN**
"CRINGENES: What is different compared to other genomic newborn screening projects?"
Judit García Villoria, Jefa de Sección - Sección Errores Congénitos del Metabolismo-IBC.BGM, Hosp Clínic, Barcelona

16h10-17h30 ▸ **COFFEE-BREAK | POSTER VIEWING AND DISCUSSION**

17h30-18h20 ▸ **SELECTED ORAL COMMUNICATIONS I**
Chairs: Ilda Ribeiro | Fábio J. Ferreira

OC 01 17h30-17h40	João Miguel Carvalho Nogueira	*Dissecting and Predicting the impact of enhancer variants in type 2 diabetes using high-throughput mutagenesis and reporter assays*
OC 02 17h40-17h50	Ana Joana Duarte	*Generation of cellular models for fabry disease: unlocking the potential of ipscs and gene editing*
OC 03 17h50-18h00	Rafael Graça	*Dissecting non-coding regulation of ldlr and PCSK9 in familial hypercholesterolaemia*
OC 04 18h00-18h10	André Besouro-Duarte	*Differential allelic expression case-control study unveils missing prostate cancer heritability and pleiotropy with breast cancer*
OC 05 18h10-18h20	Frederico Pena	*Establishment of a scalable neuronal reporter model of Machado-Joseph disease based on gene-edited patient cells as a platform for high-throughput drug repurposing*

18h30 ▸ **SPGH GENERAL ASSEMBLY**

FRIDAY 21st NOVEMBER

09h00-10h00 ▸ **SELECTED ORAL COMMUNICATIONS II**
Chairs: Jorge Saraiva | José Ferrão

OC 06 09h00-9h10	Ricardo Amorim	*Clinical trajectories and healthcare burden of hereditary diffuse gastric cancer: prevention versus treatment in european centres*
OC 07 09h10-9h20	Mariana Tomásio Neves	*Mutyh carrier frequency in portugal: navigating the data gap with what we've got*
OC 08 9h20-9h30	Cristina Santos	*Lessons learned from a portuguese inherited retinal disease research cohort*
OC 09 9h30-9h40	Ana Miguel Pinto Capela	*Understanding the genetic basis of hereditary cardiovascular diseases in portugal: a retrospective analysis across two centres*
OC 10 9h40-9h50	Sofia Quental	*RNA analysis as a first-line diagnostic approach for neurofibromatosis type 1: a six-year institutional experience*
OC 11 9h50-10h00	Filipa Oliveira	*Liquid biopsies and (EPI)genomic characterisation of glioblastoma*

10h00-11h00 ▸ **BUILDING BRIDGES:** Sociedade Portuguesa de Hematologia (in Portuguese)
"Advancing Gene Therapy for Red Blood Cell disorders – How Far Are We from an Affordable Cure"
Guests: **Tabita Magalhães Maia** (Department of Hematology, ULS Coimbra) | **José Carlos Ferreira** (Department of Obstetrics, ULS Santa Maria, Lisboa) | **Luís Pereira de Almeida**, CNC-UC & CIBB & Centro de Excelência em Terapia Genética (GeneT), Coimbra | **Salvador Payan**, Virgen del Rocío University Hospital, Institute of Biomedicine of Seville (IBiS-CSIC)
Chairs: Janet Pereira | Celeste Bento

11H00-12H00 ▸ **COFFEE-BREAK | POSTER VIEWING AND DISCUSSION**

12h00-13h00 ▸ KEYNOTE LECTURE 2

Chairs: Sérgio B. Sousa | Daniela Oliveira

12h00-12h30 ▸ *"Generative AI meets Genetics: Reasoning with GestaltMatcher"*

Peter Krawitz, University of Bonn; Institute for Genomic Statistics and Bioinformatics, Bonn, NRW, Germany

12h30-12h50 ▸ *GestaltMatcher Plenary Session: Dysmorphology Cases (TBC)*

12h50-13h10 ▸ CORPORATE SYMPOSIUM III ROCHE

"The Evolving Role of Liquid Biopsy in NSCLC: Characterizing Resistance Mechanisms"

Manuel Teixeira, Diretor do Serviço de Genética Laboratorial do IPO Porto e Coordenador do Grupo de Oncogenética do CI-IPOP

13h10-14h00 ▸ LUNCH BREAK

14h00-15h00 ▸ POSTER VIEWING AND DISCUSSION

15h00-16h00 ▸ INVITED SYMPOSIUM II: Pharmacogenomics & Precision Medicine

Pharmacogenomics and Splicing Mechanisms in Health and Disease

Chairs: Sebastião Rodrigues | Marta P. Soares

15h00-15h30 ▸ *Invited talk ISII-1: "Pharmacogenomics clinical implementation in Spanish health system: MedeA pilot project and BioFRAM"*

Adrián LLerena, INUBE Extremadura University Institute for Biosanitary Research, Faculty of Medicine and Health Sciences. Extremadura University Hospital Unit for Pharmacogenomics and Personalized Medicine. Badajoz, Spain

15h30-16h00 ▸ *Invited talk ISII-2: "Splicing tolerance and its limits in disease"*

Maria Carmo-Fonseca, Faculdade de Medicina da Universidade de Lisboa; GIMM - Gulbenkian Institute for Molecular Medicine, Lisbon, Portugal

16h00-16h30 ▸ COFFEE-BREAK

16h30-16h50 ▸ SELECTED ORAL COMMUNICATIONS III

Chairs: Henriqueta Silva | Peter Jordan

OC 12 16h30-16h40	Liliana Sousa	*Optimizing prevention and care in rare tumour risk syndromes (rtrs): insights from the preventable european consortium*
OC 13 16h40-16h50	Diogo Caetano	*Pharmacogenomic studies in medical genetics: a pilot approach to personalized medicine in practice*

17h00-17h40 ▸ ROUND TABLE: Human Genetics in Portuguese-speaking Countries (in Portuguese)

17h00-17h15 ▸ *"Molecular Research and Diagnostics in Cape Verde: Insights from Breast Cancer"*

Pâmela Borges, Molecular Biology Laboratory, Agostinho Neto University Hospital

17h15-17h40 ▸ Round table with

Raffaela Gozzelino, NOVA Medical School | Lúcio Lara Santos, IPO Porto | Isabel Carreira, FMUC-UniCV | Celeste Bento, ULS Coimbra; ALUA

17h45-18h45 ▸ INVITED SYMPOSIUM III: Hematologic Malignancies & Molecular Oncology

Chairs: Sofia Fernandes | Sara Carvalhal

17h45-18h15 ▸ *Invited talk ISIII-1: "Germline predisposition to hematological malignancies"*

Maria Julia Montoro, Vall d'Hebron University Hospitallle - Department of Haematology and Oncology, Barcelona, Spain

18h15-18h45 ▸ *Invited talk ISIII-2: "Diagnostic Experience in Bone Marrow Failure Syndromes and Hematologic Neoplasia Predisposition"*

Margarida Coucelo (Hemato-Oncology Molecular Laboratory, Department of Hematology, ULS Coimbra, Portugal)

20h00 ▸ GALA DINNER

SATURDAY 22nd NOVEMBER

08h40-09h30 ▸ SELECTED ORAL COMMUNICATIONS IV

Chairs: Ana Grangeia | Mário Laço

OC 14 08h30-08h36	Jorge Diogo Da Silva	*Early detection of juvenile myelomonocytic leukemia in CBL-related Noonan-like syndrome through exome sequencing.*
OC 15 08h36-08h42	Diana Macedo Cardoso	*The hidden risk of paracentric inversions: partial 9p trisomy and neurodevelopmental disorder*
OC 16 08h42-08h48	Daniela Oliveira	*Disruption of specc1l translation initiation by intragenic deletion: novel pathogenic mechanism in teebi-hypertelorism syndrome*
OC 17 08h48-08h54	Sara Jesus	*Somatic rhoa mosaicism unveiled by deep sequencing: dysmorphology-driven diagnosis*
OC 18 08h54-09h00	Isabel Alonso	*Integrating transcriptomics into the clinical setting for the diagnosis of sh3kbp1 deficiency*
OC 19 09h00-09h06	Pavlina Rayko	*Broadening the phenotypic spectrum of thumpd1-related syndrome: the oldest reported patient with novel features*
OC 20 09h06-09h12	Eunice Matoso	*Hells-associated episignature enables the diagnosis of ICF-4 syndrome*
OC 21 09h12-09h18	Vera Sousa Marques	*Charge syndrome with an additional FBRSL1 variant: interpreting unknown genetic findings*

09h30-10h30 ▸ BIOETHICS DEBATE: "Databases and DNA Profiles"

Comissão de Bioética (in Portuguese)

Chairs: Lina Ramos | Mariana Neves | Joaquim Sá

"Forensic DNA Profile Database: Possibilities and Limitations"

Francisco Corte Real, Instituto Nacional de Medicina Legal e Ciências Forenses

"From Biological Samples to Biobanks for Research: Ethical, Legal, and Social Aspects"

Cíntia Águas, Faculdade de Medicina, Universidade de Lisboa

10h30-11h00 ▸ CORPORATE SYMPOSIUM IV

"From Sequence to Diagnosis: Deciphering the Genome"

Daniela Piazzolla, PhD Senior Manager, Medical Affairs Europe, Illumina

"Illumina Constellation Mapped reads: A new paradigm for Whole Genome Sequencing."

Prof. Massimo Delledonne, PhD Professore di Genetica, Università di Verona

11h00-11h25 ▸ COFFEE-BREAK

11h25-12h00 ▸ KEYNOTE LECTURE 3: Metabolic Disorders & Adiposity Research

Chairs: Joana Rosmaninho-Salgado | Joana Xavier

"Causes and consequences of obesity: insights from genetics"

Sadaf Farooqi, School of Clinical Medicine, University of Cambridge, UK

12h00-12h10 ▸ SPGH IN 2026

Célia Ventura, President SPGH 2026 | Raquel Gouveia Silva, Secretary SPGH 2026, (tba), Treasurer 2026-2028

12h10-12h35 ▸ SPGH AWARD LECTURE

Chairs: (tba) | President Elected (tba)

12h35-12h55 ▸ SPGH AWARDS CEREMONY

Chair: Paula Jorge, Chair Scientific Committee

12h55-13h10 ▸ CLOSING SESSION

Sérgio B. Sousa | Janet Pereira | Cláudia Oliveira

com apoio do Município de Coimbra

CÂMARA MUNICIPAL DE COIMBRA

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