SCIENTIFIC **PROGRAMME**
PROGRAMA CIENTÍFICO

THURSDAY 20th NOVEMBER

09:00-14:00 ▸ **REGISTRATION**

10:00-12:00 ▸ **PRE-MEETING PARALLEL SESSIONS**

▸ **PROFESSIONAL**

▸ **Session 1:** *Cytogenetics and Molecular Genetics Club* (in Portuguese)
Chairs: **Isabel Carreira**, Faculty of Medicine, University of Coimbra | **Teresa Fidalgo**, Department of Hematology, ULS Coimbra

▸ **Session 2:** *Dysmorphology and Clinical Genetics Club* (in English)
Chairs: **Joana Rosmaninho-Salgado**, Department of Medical Genetics, ULS Coimbra | **Sadaf Farooqi**, School of Clinical Medicine, University of Cambridge, UK

▸ **EDUCATIONAL**

▸ **Workshop 1:**
"Genetic testing interpretation and report – beyond ACMG classification" (in Portuguese)
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" (in Portuguese)
Luís Pereira de Almeida, **Rui Nobre**, CNC-UC & CIBB & Centro de Excelência em Terapia Genética (GeneT), Coimbra, Portugal

12:10-14:00 ▸ **PRE-CONGRESS PLENARY SESSION**

12:10-12:30 ▸ **CORPORATE SYMPOSIUM I ILC**
Chair: **Cristina Iglésias da Costa**, CEO da ILC- Instrumentos de Laboratório e Científicos, lda
"Redefining Sequencing: From Genomics to 5D Multiomics with AVITI24"
Antonio Fadda, Field Applications Scientist, Element Biosciences

12:30-13:00 ▸ **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)

13:00-14:00 ▸ **LUNCH WITH PATIENT ASSOCIATIONS**

14:00-14:15 ▸ **OPENING AND WELCOME**
Sérgio B. Sousa, Janet Pereira, Cláudia Oliveira, Paula Jorge, e representantes da Câmara Municipal de Coimbra, Universidade de Coimbra e Unidade Local de Saúde de Coimbra

14:15-14:50 ▸ **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

14:50-15:50 ▸ **INVITED SYMPOSIUM I:** Gene therapy for eye disorders
Chairs: Ana Luísa Carvalho | Rui Nobre

14:50-15:20 ▸ *Invited talk ISI-1: "Gene Therapy for Inherited Retinal Diseases"*
Bart Leroy, Department of Ophthalmology, Ghent University Hospital, Ghent, Belgium

15:20-15:50 ▸ *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

15:50-16:10 ▸ **CORPORATE SYMPOSIUM II QUILABAN**
Chair: **Pedro Pinto**, Quilaban
"CRINGENES: What is different compared to other genomic newborn screening projects?"
Judit García Villoria, Sección Errores Congénitos del Metabolismo-IBC.BGM, Hosp Clínic, Barcelona

16:10-17:30 ▸ **COFFEE-BREAK | POSTER VIEWING AND DISCUSSION**

17:30-18:20 ▸ **SELECTED ORAL COMMUNICATIONS I**
Chairs: Ilda Ribeiro | Fábio J. Ferreira

OC 01 17:30-17:40	João Nogueira	*Dissecting and Predicting the impact of enhancer variants in type 2 diabetes using high-throughput mutagenesis and reporter assays*
OC 02 17:40-17:50	Ana Joana Duarte	*Generation of cellular models for fabry disease: unlocking the potential of IPSCS and gene editing*
OC 03 17:50-18:00	Rafael Graça	*Dissecting non-coding regulation of LDLR and PCSK9 in familial hypercholesterolaemia*
OC 04 18:00-18:10	André Besouro-Duarte	*Differential allelic expression case-control study unveils missing prostate cancer heritability and pleiotropy with breast cancer*
OC 05 18:10-18:20	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*

18:30 ▸ **SPGH GENERAL ASSEMBLY**

FRIDAY 21st NOVEMBER

09:00-10:00 ▸ **SELECTED ORAL COMMUNICATIONS II**
Chairs: Jorge Saraiva | José Ferrão

OC 06 09:00-09:10	Ricardo Amorim	*Clinical trajectories and healthcare burden of hereditary diffuse gastric cancer: prevention versus treatment in european centres*
OC 07 09:10-09:20	Mariana Tomásio Neves	*MUTYH carrier frequency in portugal: navigating the data gap with what we've got*
OC 08 09:20-09:30	Cristina Santos	*Lessons learned from a portuguese inherited retinal disease research cohort*
OC 09 09:30-09:40	Ana Miguel Pinto Capela	*Understanding the genetic basis of hereditary cardiovascular diseases in portugal: a retrospective analysis across two centres*
OC 10 09:40-09:50	Sofia Quental	*RNA analysis as a first-line diagnostic approach for neurofibromatosis type 1: a six-year institutional experience*
OC 11 09:50-10:00	Filipa Oliveira	*Liquid biopsies and (EPI)genomic characterisation of glioblastoma*

10:00-11:00 ▸ **BUILDING BRIDGES:** Sociedade Portuguesa de Hematologia (in Portuguese)
Chairs: Janet Pereira | Celeste Bento
"Advancing Gene Therapy for Red Blood Cell disorders – How Far Are We from an Affordable Cure"
Guests: **Salvador Payan**, Virgen del Rocío University Hospital, Institute of Biomedicine of Seville (IBiS-CSIC) | **Luís Pereira de Almeida**, CNC-UC & CIBB & Centro de Excelência em Terapia Genética (GeneT), Coimbra | **Tabita Magalhães Maia** (Department of Hematology, ULS Coimbra) | **José Carlos Ferreira** (Department of Obstetrics, ULS Santa Maria, Lisboa)

11:00-12:00 ▸ **COFFEE-BREAK | POSTER VIEWING AND DISCUSSION**

12:00-13:00 ▸ **KEYNOTE LECTURE 2**
Chairs: Sérgio B. Sousa | Daniela Oliveira
12:00-12:30 ▸ *"Generative AI meets Genetics: Reasoning with GestaltMatcher"*
Peter Krawitz, University of Bonn; Institute for Genomic Statistics and Bioinformatics, Bonn, NRW, Germany
12:30-12:50 ▸ *GestaltMatcher in practice: Portuguese clinical cases*

12:50-13:10 ▸ **CORPORATE SYMPOSIUM III ROCHE**
Chair: **Catarina Salgado**, Medical Manager, 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, Porto

13:10-14:00 ▸ **LUNCH BREAK**

14:00-15:00 ▸ **POSTER VIEWING AND DISCUSSION**

15:00-16:00 ▸ **INVITED SYMPOSIUM II:** Pharmacogenomics & Precision Medicine
Pharmacogenomics and Splicing Mechanisms in Health and Disease
Chairs: Sebastião Rodrigues | Marta P. Soares
15:00-15:30 ▸ *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
15:30-16:00 ▸ *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

16:00-16:30 ▸ **COFFEE-BREAK**

16:30-16:50 ▸ **SELECTED ORAL COMMUNICATIONS III**
Chairs: Henriqueta Silva | Peter Jordan

OC 12 16:30-16:40	Liliana Sousa	*Optimizing prevention and care in rare tumour risk syndromes (RTRS): insights from the preventable european consortium*
OC 13 16:40-16:50	Diogo Caetano	*Pharmacogenomic studies in medical genetics: a pilot approach to personalized medicine in practice*

17:00-17:40 ▸ **ROUND TABLE:** Human Genetics in Portuguese-speaking Countries - Cabo Verde (in Portuguese)
17:00-17:15 ▸ *"Molecular Research and Diagnostics in Cape Verde: Insights from Breast Cancer"*
Pâmela Borges, Molecular Biology Laboratory, Agostinho Neto University Hospital, Praia, Cape Verde
17:15-17:40 ▸ Round table with
Raffaella Gozzelino, NOVA Medical School | **Lúcio Lara Santos**, IPO Porto | **Isabel Carreira**, FMUC-UniCV | **Celeste Bento**, ULS Coimbra; ALUA

17:45-18:45 ▸ **INVITED SYMPOSIUM III:** Hematologic Malignancies & Molecular Oncology
Chairs: Sofia Fernandes | Sara Carvalhal

17:45-18:15 ▸ *Invited talk ISIII-1: "Germline predisposition to hematological malignancies"*
Maria Julia Montoro, Vall d'Hebron University Hospitallle - Department of Haematology and Oncology, Barcelona, Spain

18:15-18:45 ▸ *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

20:00 ▸ **CONFERENCE DINNER**

SATURDAY 22nd NOVEMBER

08:30-09:30 ▸ **SELECTED ORAL COMMUNICATIONS IV - Clinical Cases**
Chairs: Ana Grangeia | Mário Laço

OC 14 08:30-08:36	Jorge Diogo Da Silva	*Early detection of juvenile myelomonocytic leukemia in CBL-related Noonan-like syndrome through exome sequencing.*
OC 15 08:36-08:42	Diana Macedo Cardoso	*The hidden risk of paracentric inversions: partial 9p trisomy and neurodevelopmental disorder*
OC 16 08:42-08:48	Daniela Oliveira	*Disruption of SPECC1L translation initiation by intragenic deletion: novel pathogenic mechanism in teebi-hypertelorism syndrome*
OC 17 08:48-08:54	Sara Jesus	*Somatic RHOA mosaicism unveiled by deep sequencing: dysmorphology-driven diagnosis*
OC 18 08:54-09:00	Isabel Alonso	*Integrating transcriptomics into the clinical setting for the diagnosis of SH3KBP1 deficiency*
OC 19 09:00-09:06	Pavlina Rayko	*Broadening the phenotypic spectrum of THUMPD1-related syndrome: the oldest reported patient with novel features*
OC 20 09:06-09:12	Eunice Matoso	*Hells-associated episignature enables the diagnosis of ICF-4 syndrome*
OC 21 09:12-09:18	Vera Sousa Marques	*CHARGE syndrome with an additional FBRSL1 variant: interpreting unknown genetic findings*

09:30-10:30 ▸ **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

10:30-10:50 ▸ **CORPORATE SYMPOSIUM IV ILLUMINA**
"From Sequence to Diagnosis: Deciphering the Genome"
Chair: **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

10:50-11:20 ▸ **COFFEE-BREAK**

11:20-11:55 ▸ **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

11:55-12:05 ▸ **SPGH IN 2026**
Célia Ventura, President SPGH 2026 | **Raquel Gouveia Silva**, Secretary SPGH 2026, Treasurer 2026-2028 (tba)

12:05-12:30 ▸ **SPGH AWARD LECTURE**
Chairs: **Sebastião Rodrigues**, Co-chair Scientific Committe | **President Elected (tba)**

12:30-12:50 ▸ **SPGH AWARDS CEREMONY**
Chair: **Paula Jorge**, Chair Scientific Committee

12:50-13:00 ▸ **CLOSING SESSION**
Sérgio B. Sousa | Janet Pereira | Cláudia Oliveira