28TH ANNUAL MEETING

SOCIEDADE PORTUGUESA DE GENÉTICA HUMANA

5 - 7 DECEMBER 2024

INSTITUTO SUPERIOR DE ENGENHARIA
ISEP PORTO

Rua António Bernardino Almeida, 431
4200-529 Porto

SPGH
Sociedade Portuguesa de Genética Humana

Programme

Thursday 5th December

09h00-14h00 › Registration

10h00-12h00 › PRE-MEETING CONCURRENT SESSIONS

› **EDUCATIONAL SESSION 1:** *Scientific Writing*
Helder Maiato – *i3S, Porto*

› **CLUB 1:** *Cytogenetics and Molecular Genetics – Case discussion*
Sofia Dória – *FMUP, Porto* | **Natália Salgueiro – *Synlab, Porto***

› **EDUCATIONAL SESSION 2:** *Classification of variants – Rules and criteria*
Sónia Sousa | Sofia Quental – *Ipatimup Diagnósticos, Porto*

› **CLUB 2:** *Dysmorphology and Clinical Genetics*
Pedro Louro – *ULSSJ, Porto* | **Ana Rita Soares – *ULSSA, Porto***

12h30-13h30 › CORPORATE MEETINGS (concurrent in 2 rooms)

› **QUILABAN -** "*Unlocking genomic insights for rare and hereditary disease with Emedgene*", **Duarte Oliveira**

› **LIMBUS MEDICAL TECHNOLOGY -** "*Exome diagnostics and beyond: How to navigate the NGS universe with confidence using the varvis® software*"

Chair: **Bruno Pescara**
Part 1: *How standardized filtering strategies accelerate exome diagnostics,* **Ximena Escalera-Fanjul**
Part 2: *Cabinet of curiosities: Detecting and interpreting extraordinary variants*, **Ben Liesfeld**

13h30-14h00 › Lunch break – *Lunch offered to attendees of corporate meetings*

14H00-14h15 › OPENING AND WELCOME

Carla Oliveira | President SPGH 2024 **& Sebastião Rodrigues** | Chair Scientific Committee

14h15-14h50 › KEYNOTE LECTURE 1

Chair: **Peter Jordan | Sebastião Rodrigues**
"*Single cell human genetics: understanding human disease one cell at a time*"
Malte Spielmann *(Institute of Human Genetics, Lubeck, Germany)*

14h50-15h50 › Invited Symposium I (IS-I): *Genetics of Neurological Disorders*

Chair: **João Paulo Oliveira | Jorge Oliveira**

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): AstraZeneca/MSD

"*Genetic Testing and PARP Inhibitors in Portugal: assessing the present and charting the course forward*"

Breast cancer | Cláudia Vieira *(IPO Porto)*

Prostate cancer | Ana Faria *(ULS Loures Odivelas)*

16h10-17h30 › *Coffee-break* | Poster viewing and Discussion | Session I

17h30-18h20 › SELECTED ORAL PRESENTATIONS I: *Basic Research*

Chair: Bibiana Ferreira | João Vinagre

17h30-17h40	Ana Raquel Soares	tRNA MODIFICATIONS ARE POTENTIAL ALZHEIMER'S DISEASE BIOMARKERS AND THERAPEUTIC TARGETS THAT CAN BE REPROGRAMMED THROUGH tRNA MODIFYING ENZYME EXPRESSION MANIPULATION TO RECOVER CELLULAR FITNESS
17h40-17h50	Ana Catarina Nunes	GENOME.WIDE CRISPR SCREEN IDENTIFIES RAB14 AS A NEW TARGET FOR GASTRIC CANCER TREATMENT
17h50-18h00	Susana Valente	RUNS OF HOMOZYGOSITY: BIOINFORMATIC APPROACHES FOR DIAGNOSTIC PURPOSES AND POPULATION ANALYSIS USING A SAMPLE OF 12,167 EXOMES
18h00-18h10	Liliana Matos	A 5' SPLICE-STE MUTATION CAUSING MUCOLIPIDOSIS TYPE III CAN BE EFFICIENTLY RESCUED BY U1 snRNA-BASED THERAPY
18h10-18h20	Maria João Pinho	THE IMPACT OF BALANCED CHROMOSOMAL REARRANGEMENTS IN PGT-SR

18h30 › SPGH GENERAL ASSEMBLY

Friday 6th December

09H00-10h00 › SELECTED ORAL PRESENTATIONS II: *Clinical research*

Chair: Célia Soares | Ana Luísa Carvalho

09h00-09h10	Luís Nunes	CO-OCCURRING MUTATIONS IDENTIFY PROGNOSTIC SUBGROUPS OF MICROSATELLITE STABEL COLRECTAL CANCER
09h10-09h20	José Garcia-Pelaez	CLINICAL CRITERIA FOR CDH1 GERMLINE TESTING ACROSS DIFFERENT WORLD REGIONS: SHALL WE KEEP THEM GLOBAL?
09h20-09h30	Filipe Alves	CLINICAL EXOME CONCEPTUALIZATION: A BIOINFORMATICS FRAMEWORK FOR SEMI-AUTOMATED REVIEW OF GENE-PHENOTYPE ASSOCIATIONS
09h30-09h40	Domingos Roda	CLINICAL IMPACT OF DYNAMIC CHANGES IN CFDNA DURING CHEMO/RADIATION IN GLIOBLASTOMA PATIENTS
09h40-09h50	André M. Travessa	IS MILD BONE FRAGILITY GENETIC? - GENETIC FINDINGS IN A COHORT OF PATIENTS WITH EARLY-ONSET FRACTURES AND/OR OSTEOPOROSIS AND NO EXTRASKELETAL SIGNS OF OSTEOGENESIS IMPERFECTA

10h00-11h00 › BUILDING BRIDGES: *Hereditary Cancer Network in Portugal*

Chair:Carla Oliveira

Guests: Carla Pereira (*Department of Quality in Health, Ministry of Health of Portugal*) | **Fátima Vaz** (*ERN Genturis Reference Centre IPOL*) | **Manuel Teixeira** (*ERN Genturis Reference Centre IPOP*) | **Luzia Garrido** (*ERN Genturis Reference Centre ULSSJ*)

11h00-12h00 › Coffee-break / Poster Viewing and Discussion, Session II

12h00-13h00 › INVITED SYMPOSIUM II (ISII): *Update on preimplantation genetic testing and prenatal diagnosis in Portugal*

Chair: Ana Grangeia | Rosário Pinto Leite

12h00-12h30 › INVITED TALK ISII-1: "*Preimplantation genetic testing in Portugal*"

Filipa Carvalho *(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 III**

15h00-16h00 › **Invited Symposium III (ISIII):** *Cancer Genetics*

Chair: **Joana Xavier | Sofia Fernandes**

15h00-15h30 › **Invited talk ISIII-1:** "*Genetics of Li-Fraumeni*"

Gaëlle Bougeard *(Universitè 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: **Marisa Silva | Joana Salgado**

16h30-16h40	Luís Miguel Pires	CLINICAL UTILITY OF DNA METHYLATION EPISIGNATURES TESTING - A PORTUGUESE MULTICENTER COHORT STUDY
16h40-16h50	Rita Barbosa-Matos	PERSONALIZED REGION-SPECIFIC LIFETIME CANCER RISK ESTIMATES FOR AN ANCESTRAL CDH1 VARIANT

16h30-16h50 › **ROUND TABLE:** *Bridging imaging and genetics*

Chair: **André Travessa | Marta Soares**

João Soares Fernandes *(Hospital de Braga, Braga, Portugal)*

José Almeida *(Champalimaud Foundation, Lisbon, Portugal)*

17h45-18h45 › **Invited Symposium IV (ISIV):** *Understanding and treating disease*

Chair: **Margarida Venâncio | Ilda Ribeiro**

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 December

08H40-09h30 › **SELECTED ORAL PRESENTATIONS IV:** *Clinical cases*

Chair: **Ana Raquel Silva | Susana Fernandes**

08h40-08h47	Sara Pinho	EXPANDING THE PHENOTYPE ASSOCIATED WITH IRF2BPL GENE VARIANTS: A REPORT OF 4 NEW CASES
08h47-08h54	Catarina Macedo	PRENATAL AND POSTNATAL PHENOTYPING AND MOLECULAR CHARACTERIZATION OF THREE CASES OF ZTTK SYNDROME: A POTENTIALLY RECOGNIZABLE CLINICAL ENTITY
08h54-09h01	Ana Miguel Capela	CTCF-RELATED INTELLECTUAL DISABILITY – FOUR PATIENTS SUGGESTING A RECOGNIZABLE FACIAL GESTALT
09h01-09h08	Isabel Serra Nunes	FRAXE INTELLECTUAL DEVELOPMENT DISORDER – WARNING FOR A FORGOTTEN DIAGNOSIS
09h08-09h15	Nuno Teixeira	A NEVER-ENDING RACE: A MULTILOCUS CAUSE OF AN INBORN ERROR OF IMMUNITY ASSOCIATED WITH COMPLEX NEUROLOGICAL DISEASELETAL SIGNS OF OSTEOGENESIS IMPERFECTA
09h15-09h22	Lígia Lameiras	A CHALLENGING DETECTION OF A NOVEL SPLICING DELETION IN THE DMD GENE IN A PATIENT WITH DUCHENNE MUSCULAR DYSTROPHYSKELETAL SIGNS OF OSTEOGENESIS IMPERFECTA
09H22-09H29	Sofia Fragoso	BEYOND THE AFFECTED: THE IMPORTANCE OF TESTING NON-AFFECTED INDEX INDIVIDUALS IN CANCER HIGH-RISK FAMILIES

09h30-10h30 › BIOETHICS DEBATE: "*Lack of access to healthcare in rare diseases*"

Chair: **Lina Ramos | Joaquim Sá**

"European point of view" **Dorica Dan** *(EURORDIS/ePAG)*

"A Portuguese approach" **Mariana Neves** *(Serviço Genética Médica, ULSSM)*

10h30-11h00 › CORPORATE SYMPOSIUM II (CSII) – QUILABAN | Illumina

"Non-invasive prenatal testing for fetal aneuploidies by Cell-Free DNA whole-genome sequencing"

Luís Miguel Pires and Oscar Bolanos

11h00-11h25 › Coffee-break

11h25-12h00 › KEYNOTE LECTURE 2

Chair: **Sérgio Sousa | Celeste Bento**

"Genoprotective interventions for healthspan extension"

Elsa Logarinho *(i3S, Porto, Portugal)*

12h00-12h10 › SPGH in 2025 | **Sérgio Sousa,** *President SPGH 2025* | **Janet Pereira, Cláudia Oliveira**

12h10-12h35 › SPGH Award Lecture

Chair: **Sebastião Rodrigues** | President Elected (tba)

12h35-12h55 › SPGH Awards Ceremony

Chair: **Sebastião Rodrigues | Paula Jorge**

12h55-13h10 › Closing Session

Carla Oliveira | Ana Grangeia | Cláudia Oliveira