SPGH

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09:00 **OPENING & WELCOME**

09:15 **25TH ANNIVERSARY SESSION**

Chairs: Miguel Guimarães (Bastonário OM), Carla Oliveira

AMÂNDIO S. TAVARES AND THE FOUNDATIONS OF HUMAN GENETICS IN PORTO

Jorge Sequeiros (i3S, Porto,PT)

THE BEGINNINGS OF MEDICAL GENETICS AND THE CREATION OF SPGH - CONTRIBUTION TO THE HISTORY OF ITS FOUNDATION

Heloísa Santos, (1st President of the SPGH, PT)

KEYNOTE: DNA METHYLATION LANDSCAPES OF BREAST CANCER REVEAL EPIGENETIC INSTABILITY AND CIS REGULATION

Carlos Caldas, (CRUK Cambridge Institute, Cambridge, UK)

10:15 COFFEE-BREAK / ONLINE EXHIBITION

10:25 **INVITED SYMPOSIUM I (IS-I):**

CHROMATIN REMODELLING IN HEALTH AND DISEASE

Chairs: Sérgio Sousa, Sebastião Rodrigues

ISI-1: THE PANCREAS REGULOME: FROM CAUSALITY TO PREDICTION OF NON-CODING MUTATIONS IN HUMAN PANCREATIC DISEASES

José Bessa (i3S, Porto, PT)

ISI-2: ROLE OF BAF SWI/SNF CHROMATIN REMODELLERS IN NEURODEVELOPMENT AND ID

Cristina Dias (CRICK, London, UK)

11:25 **CORPORATE SYMPOSIUM**
PRENATAL TRIO EXOME SEQUENCING FOR FETAL ULTRASOUND ANOMALIES
Detlef Trost

11:45 **CORPORATE SYMPOSIUM**
GENE THERAPY – FROM GENESIS TO TREATMENT

12:25 **CORPORATE SYMPOSIUM**
GENETIC DIAGNOSIS OF GASTROINTESTINAL HEREDITARY CANCER SYNDROMES: NGS UPDATE
Cristina Albuquerque

12:45 LUNCH BREAK

13:10 **POSTER VIEWING AND DISCUSSION / ONLINE EXHIBITION**

14:40 **SELECTED ORAL PRESENTATIONS I - BASIC RESEARCH**
Chairs: Joana Melo, Paula Jorge

OPI_1 - MUTANT ATAXIN-2 PATHOLOGICAL FEATURES IS ALTERED BY THE AGING PROCESS
Inês Afonso | Faculdade de Medicina e Ciências Biomédicas, Universidade do Algarve, Faro, Portugal, Algarve Biomedical Center – Research Institute, Faro, Portugal

OPI_2 - THE CDH1 LOCUS REGULATORY ARCHITECTURE: CDH1 NONCODING ELEMENTS CONTROL E-CADHERIN CANONICAL FUNCTIONS
Celina São José | i3S/Ipatimup, Porto, Portugal; FMUP, Porto, Portugal; MPIMG, Berlin, Germany

OPI_3 - TRANSCRIPTOMIC CHARACTERIZATION OF HUMAN IPSC-DERIVED CARDIOMYOCYTES
Beatriz Gomes Silva | Instituto de Medicina Molecular

OPI_4 - DEFINING THE EVOLUTIONARY BASIS OF MALE GERM CELL IDENTITY: AN INTEGRATED BASIC AND CLINICAL RESEARCH APPROACH
Paulo Navarro-Costa | Instituto Gulbenkian de Ciência

OPI_5 - WHOLE GENOME SEQUENCING ANALYSIS: EXPLORING GERMLINE CNV LANDSCAPES
Marta Ferreira | i3S/ipatimup, Porto, Portugal; FCUP, Porto, Portugal

15:55 **PANEL DISCUSSION (PDI):**

EMERGENT CHALLENGES IN HUMAN GENETICS

Chairs: Ana Berta Sousa, Jorge Saraiva

PDI-1: GENETIC CARRIER SCREENING IN PORTUGAL: TOWARDS A SCIENTIFICALLY SOUND, ETHICALLY RIGOROUS, SOCIALLY RESPONSIBLE PROGRAMME
Jorge Sequeiros (i3S, Porto, PT)

PDI-2: TECHNICAL STANDARDS OF THE INTERPRETATION AND REPORTING OF CONSTITUTIONAL COPY-NUMBER VARIANTS
Erin Rooney Riggs (USA)

16:55 COFFEE-BREAK / POSTER VIEWING AND DISCUSSION / ONLINE EXHIBITION

17:10 **SELECTED ORAL PRESENTATIONS II - CLINICAL RESEARCH**
Chairs: Sofia Dória, Gabriel Miltényi-Miltenberger

OPII_1 - GERMLINE COPY NUMBER VARIANTS: AN UNDERREPORTED GENETIC DIAGNOSIS IN GASTROINTESTINAL TUMOUR RISK SYNDROME SUSPECTED INDIVIDUALS
José García-Peláez | IPATIMUP/i3S

OPII_2 - HEAD AND NECK SQUAMOUS CELL CARCINOMA SIGNATURES: AN INTEGRATIVEMULTI-OMICS APPROACH
Luísa Esteves | University of Coimbra, Cytogenetics and Genomics Laboratory, Institute of Cellular andMolecular Biology, Faculty of Medicine, Coimbra, Portugal

OPII_3 - INCIDENTAL CARRIER DETECTION OF 639 VARIANTS IN PATIENTS TESTED FORDIAGNOSTIC PURPOSES: ADDED VALUE FOR GENETIC COUNSELLING AND A GLIMPSEABOUT RECESSIVE DISEASES IN PORTUGAL
Fátima Lopes | CGPP - Centro de Genética Preditiva e Preventiva, Porto, Portugal, IBMC – Instituto deBiologia Molecular e Celular, Universidade do Porto, Porto, Portugal

OPII_4 - THE FIRST CDH1 FOUNDER VARIANT IN THE PORTUGUESE POPULATION: A MISSENSEWITH SEVERE IMPACT IN MRNA SPLICING
Rita Barbosa-Matos | i3S; Ipatimup; Doctoral Programme on Cellular and Molecular Biotechnology Applied toHealth Sciences

OPII_5 - DISCLOSURE OF GENETIC INFORMATION TO PATIENTS' RELATIVES: HEALTHCAREPROFESSIONALS' PERSPECTIVE ON PERCEIVED RESPONSIBILITIES ANDCONFIDENTIALITY OF GENETIC INFORMATION
Álvaro Mendes | IBMC, i3S

18:25 GENERAL ASSEMBLY

08:30 **SELECTED ORAL PRESENTATIONS II - CLINICAL CASE REPORTS**

Chairs: Gabriela Soares, Lina Ramos

OPIII_1 - TRICHOTHIODYSTROPHY, A CASE-REPORT HIGHLIGHTING THE IMPORTANCE OFCONNECTING GENOTYPE AND PHENOTYPE DATA TO REACH A DIAGNOSIS
Ana M. Capela | Unidade de Genética Médica, Centro de Genética Médica Jacinto Magalhães – Centro Hospitalar Universitário do Porto, Portugal

OPIII_2 - HEART DEFECTS, ORAL CLEFT, AND POLYDACTYLY CAUSED BY BIALLELIC VARIANTS INWDPCP GENE INVOLVED IN CILIOGENESIS
Rita Quental | Serviço de Genética Médica, Centro Hospitalar Universitário de São João, Porto, Portugal

OPIII_3 - MANDIBULOFACIAL DYSOSTOSIS TYPE GUION-ALMEIDA: NATIONAL CASE SERIES WITHCLINICAL AND MOLECULAR CHARACTERIZATION
Mafalda Melo | Serviço de Genética Médica, Hospital Dona Estefânia, Centro Hospitalar Universitário de Lisboa Central, Lisboa, Portugal

OPIII_4 - THREE PATIENTS WITH PHIP-RELATED SYNDROME - FURTHER PHENOTYPIC DELINEATION
Catarina Macedo | Serviço de Genética Médica, Departamento de Pediatria, Hospital de Santa Maria, Centro Hospitalar e Universitário Lisboa Norte, Lisboa, Portugal

OPIII_5 - EXPANDING THE CLINICAL SPECTRUM OF COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY ASSOCIATED WITH MRPS34 GENE: A MILD CASE WITH SLENDER HABITUS
André M. Travessa | Serviço de Genética Médica, Departamento de Pediatria, Hospital de Santa Maria, Centro Hospitalar Universitário Lisboa Norte, Lisboa, Portugal

09:30 **BIOETHICS DEBATE (BE): ETHICAL IMPERATIVE – THE IMPORTANCE OF QUALITY ON DIAGNOSTICS AND PRACTICE IN GENETIC MEDICINE**

Chairs: Célia Ventura, Carolino Monteiro

BE-1: THE CLINICAL GENETICIST PERSPECTIVE
Lina Ramos (CHUC, Coimbra, PT)

BE-2: THE LABORATORY GENETICIST PERSPECTIVE
Belinda Xavier (CHUV, Lausanne, CH)

BE-3: THE GENETICS SCIENTIST PERSPECTIVE
Carla Oliveira (i3S/Ipatimup, Porto, PT)

GENERAL DISCUSSION
Heloísa Santos, (1st President of the SPGH (PT))

10:30 COFFEE-BREAK / ONLINE EXHIBITION

10:45 **CORPORATE SYMPOSIUM**
A NEW TREATMENT FOR CHILDREN WITH ACHONDROPLASIA
Melita Irving (London/UK)

11:05 **CORPORATE SYMPOSIUM**
ADVANCES IN WGS APPLICATIONS: LATEST COLLABORATIONS AND INNOVATIONS
Martin Boura

11:45 **CORPORATE SYMPOSIUM**
DISTROFIAS DA RETINA: A ATUALIDADE DO DIAGNÓSTICO E TRATAMENTO, E PERSPETIVAS FUTURAS
Ana Luísa Carvalho (CHUC/Coimbra/PT)

12:05 LUNCH BREAK

12:30 **POSTER VIEWING AND DISCUSSION / ONLINE EXHIBITION**

14:00 **INVITED SYMPOSIUM II (ISII):**
STUDY MODELS AND TREATMENTS IN GENETIC DISEASES
Chair: Carla Oliveira

ISII-1: SPINAL MUSCULAR ATROPHY TREATMENT UPDATES
Eduardo Tizzano (VHIO, Barcelona, ES)

ISII-2: NEUROFIBROMATOSIS TREATMENT UPDATES
João Passos (IPOL, Lisboa, PT)

ISII-2: SYNTHETIC LETHALITY: PROMISES AND HURDLES IN CANCER TREATMENT
Chris Lord (CRUK, London, UK)

15:25 **KEYNOTE LECTURE**

Chair: João Gonçalves

TELOMERE MAINTENANCE, CHROMOSOMAL INTEGRITY AND THE SPECTRUM OF HUMAN DISEASES"

Luis Batista (Siteman Cancer Center, Washington, USA)

16:00 COFFEE-BREAK / POSTER VIEWING AND DISCUSSION / ONLINE EXHIBITION

16:20 **NEW EUROPEAN PROJECTS IN HUMAN GENETICS**

Chair: Carla Oliveira

16:50 **SPGH AWARDS CEREMONY**

17:15 **CLOSING SESSION**

SPGH

Sociedade Portuguesa
de Genética Humana

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