Centro de Congressos do Instituto Superior Técnico

Thursday, 23rd Nov

10h00-12h00 – **SPGH CLUB MEETINGS (concurrent)**

› **CLUB 1: Cytogenetic and Molecular Genetics Club**

Chair: *Hildeberto Correia and João Gonçalves*

› **CLUB 2: Dysmorphology and Clinical Genetics Club**

Chair: *Margarida Venâncio and Ana Berta Sousa*

12h00-14h00 – **REGISTRATION**

13h45-14h00 – **OPENING & WELCOME**

Peter Jordan, José Carlos Ferreira, Cláudia Oliveira, Sebastião Rodrigues

14h00-15h00 – **KEYNOTE LECTURE I**

Chair: *Carla Oliveira; Peter Jordan*

Solving the Unsolved Rare Diseases.

Alex Hoischen, Radboud University Medical Centre, Nijmegen, Netherlands

15h00-16h00 – **INVITED SYMPOSIUM I (ISI): GENETIC PLAYERS IN PSYCHIATRIC CONDITIONS**

Chair: *Joana B. Melo; Sérgio Sousa*

15h00-15h30 – **ISI-1: Role of psychiatric risk genes in learning and memory**. *Jeremy Hall,* Division of Psychiatry and Clinical Neuroscience, Neuroscience and Mental Health Innovation Institute, Cardiff University

15h30-16h00 – **ISI-2: Genotype-phenotype correlations in Neurodevelopmental Disorders**. *Miguel Castelo-Branco*, Coimbra Institute for Biomedical Imaging and Translational Research

16h00-16h20 – **CORPORATE SYMPOSIUM I › BIONANO**

Revolutionizing Cytogenomics with Optical Genome Mapping: Sample-to-answer workflow. Now powered by VIA software. *Alicia Bertolotti*, Bionano Genomics

16h20-17h00 – Coffee-break / Poster viewing

17h00-18h00 – **SELECTED ORAL PRESENTATIONS I (BASIC RESEARCH)**

Chair: *Clévio Nóbrega; Sebastião Rodrigues*

	Title	Author
OPI-1 17h00-17h10	TOWARDS PERSONALIZED THERAPEUTICS FOR NEURODEVELOPMENTAL DISORDERS: A STEM-CELL BASED DISEASE MODEL OF ANGELMAN SYNDROME	*Evguenia Bekman*
OPI-2 17h10-17h20	DISSECTING THE ROLE OF MICRORNAS IN EFFECTOR VERSUS REGULATORY CD4+ T CELL DIFFERENTIATION DURING (AUTO)IMMUNE RESPONSES IN VIVO	*Anita Q. Gomes*
OPI-3 17h20-17h30	THE CDH1 REGULATORY BLUEPRINT: SHAPING HEREDITARY DIFFUSE GASTRIC CANCER	*Celina São José*
OPI-4 17h30-17h40	USING HRDETECT TO TUNE THE CLASSIFICATION OF VARIANTS OF UNCERTAIN SIGNIFICANCE IN A PAN-CANCER COHORT OF 10,675 SAMPLES	*Orlando Pimenta Rodrigues*
OPI-5 17h40-17h50	A PROMISING SUBSTRATE REDUCTION APPROACH FOR A RARE GENETIC DISEASE: MUCOPOLYSACCHARIDOSES TYPE III	*Juliana I. Santos*

18h15-19h30 – **SPGH General Assembly**

Friday, 24th Nov

09h00-10h00 – **SELECTED ORAL PRESENTATIONS II (Clinical Research)**

Chair: *Inês Carvalho; Hugo Pinheiro*

	Title	Author
OPII-1 09h00-09h10	THE SOMATIC LANDSCAPE OF DIFFUSE GASTRIC CANCERS OCCURRING IN CTNNA1 GERMLINE VARIANT CARRIER PATIENTS	*Silvana Lobo*
OPII-2 09h10-09h20	MEDICALLY ACTIONABLE SECONDARY FINDINGS FROM WHOLE EXOME SEQUENCING (WES) DATA IN A SAMPLE OF 3,972 INDIVIDUALS	*Mafalda Melo*
OPII-3 09h20-09h30	WHOLE-EXOME SEQUENCING AT A PORTUGUESE MEDICAL GENETICS CENTRE: 883 CASES OVER AN 8-YEAR PERIOD	*Sara Pinho*
OPII-4 09h30-09h40	HICKAM'S DICTUM: KEEPING UP WITH THE DIAGNOSES	*Susana Lemos Ferreira*
OPII-5 09h40-09h50	AN INSIGHT ON THE ORIGINS OF HEREDITARY DIFFUSE GASTRIC CANCER THROUGH ORGANOID DRIVEN ORGAN ON-A-CHIP TECHNOLOGY	*Daniel A Ferreira*

10h00-11h00 – **BUILDING BRIDGES**: RASTREIO NEONATAL NACIONAL. QUE FUTURO?

SPGH – Sociedade Portugeusa de Genética Humana**.** *Joaquim Sá*

SPN – Sociedade Portuguesa de Neonatologia. *Inês Girbal*

PVNPC – Programa de Vigilância Nacional da Paralisia Cerebral. *Eulália Calado*

SPDM – Sociedade Portuguesa de Doenças Metabólicas**.** *Rita Jotta*

Chair: *Lina Ramos*

11h00-11h20 – Coffee-break

11h20-11h40 – SELECTED ORAL PRESENTATIONS III (Clinical Cohorts)

Chair: *Paula Jorge; Rosário Pinto Leite*

	Title	Author
OPIII-1 11h20-11h30	CHARACTERIZING HERITABLE TP53-RELATED SYNDROME IN PORTUGAL: INSIGHTS FROM THE FIRST NATIONAL COHORT	*Rita Quental*
OPIII-2 11h30-11h40	CLINICAL AND MOLECULAR CHARACTERISATION OF BAF COMPLEX-RELATED SYNDROMES IN THE PORTUGUESE POPULATION	*Daniela Oliveira*

11h40-12h40 – **INVITED SYMPOSIUM II (ISII): CANCER GENOMICS**

Chair: *João Vinagre; Mariana Soeiro e Sá*

11h40-12h10 – **ISII-1: Recent Discoveries in the Genetics of Familial Colorectal Cancer and Polyposis**. *Laura Valle,* Catalan Institute of Oncology, 08908 Barcelona, Spain

12h10-12h40 – **ISII-2: Prognostic and predictive biomarkers in colorectal cancer: lessons from Scandinavian population-based cohorts**. *Luís Nunes,* Department of Immunology, Genetics and Pathology, Science for Life Laboratory, Uppsala University

12h40-13h00 – **CORPORATE SYMPOSIUM II › ASTRAZENECA**

Pesquisa BRCA1/2 em Cancro da Mama Precoce no contexto de decisão terapêutica: impacto na prática clínica. *Ana Berta Sousa e Juliette Dupont*, Serviço de Genética do Centro Hospitalar e Universitário de Lisboa Norte

13h00-14h00 – **Lunch Break**

14h00-15h30 – **POSTER VIEWING AND DISCUSSION**

15h30-15h50 – **CORPORATE SYMPOSIUM III › ILC**

Avity in action: Comparative data for TWIST Bioscience WES in house protocol.
Natália Salgueiro, Coordenadora da Unidade de Genética Molecular e Genómica da Synlabhealth Genética Médica

15h50-16h50 – **INVITED SYMPOSIUM III (ISIII): LESSONS FROM GENETIC MODELS – THERAPY APPLICATION**
"New models for Genetic diseases and therapies"

Chair: *Ana Luísa Carvalho; Dulce Quelhas*

15h50-16h20 – **ISIII-1: "ZEBRAFISH AVATARS" predict response to chemotherapy in colorectal cancer and breast cancer-towards personalized medicine.**
Rita Fior, Champalimaud Center for the Unknown, Lisbon

16h20-16h50 – **ISIII-2**: **Molecular Therapies for Inherited Retinal and Neurometabolic Diseases.**
Alex Garanto, Radboud University Medical Center, Nijmegen, NL

16h50-17h15 – Coffee-break

17h15-17h40 – **CORPORATE SYMPOSIUM V › QUILABAN**

Illumina: Integrated Solutions to Empower Genetic Disease Testing.
Aly Villasenor PhD., Sr. Reg. Segment Marketing Manager, Genetic Disease Testing, Europe, Illumina, and
Duarte Oliveira PhD., Staff Clinical Field Application Scientist, Reproductive Health and Rare Disease, Europe, Illumina

17h40-18h40 – **KEYNOTE LECTURE II**

Chair: *Gabriela Soares; Ilda Ribeiro*

Epigenetics in Tumorigenesis and Inheritance.

Eric Lieberman Greer, Harvard Medical School

20h00 – CONGRESS DINNER

Saturday, 25th Nov

09h00-10h00 – **SELECTED ORAL PRESENTATIONS IV** (Clinical Cases)

Chair: *Ana Grangeia; Natalia Tkachenko*

	Title	Author
OPIV-1 09h00-09h10	A NOVEL FRAMESHIFT VARIANT IN NADSYN1 GENE ASSOCIATED WITH VERTEBRAL, CARDIAC, RENAL, AND LIMB DEFECTS SYNDROME 3	*Marisa Rodrigues*
OPIV-2 09h10-09h20	HIGH-LEVEL OF GNA11 MOSAICISM IN A CASE OF SYNDROMIC CUTIS MARMORATA TELANGIECTATICA CONGENITA– A CASE REPORT COMPARED WITH THE LITERATURE	*Mariana Neves*
OPIV-3 09h20-09h30	FIRST FAMILIAL CASE OF 16P13.3 MICRODELETION SYNDROME – TWO NEW CASES ILLUSTRATING THE PHENOTYPE OF THIS RECENTLY DESCRIBED SYNDROME	*C. Macedo*
OPIV-4 09h30-09h40	POSSIBILITY OF PRENATAL DIAGNOSIS IN THE OFFSPRING OF HEALTHY ROMANI COUPLES: TWO RECESSIVE PATHOLOGIES, THREE CASES, FOUR DIAGNOSES	*Gonçalo Aragão*
OPIV-5 09h40-09h50	UNEXPLAINED CARDIORESPIRATORY ARREST IN PEDIATRIC AGE: POSTMORTEM DIAGNOSIS OF ARRHYTHMOGENIC CARDIOMYOPATHY	*Catarina F. Silva*

10h00-11h00 – **BIOETHICS ROUND TABLE: New screenings, ethical reflections and recommendations**

Heloísa Santos, President of the Bioethics Committee.
Includes: "Polygenic (risk) scores in Preimplantation Genetic Testing"
Francesca Forzano, Clinical Genetics Department, Guy's and St Thomas NHS Foundation Trust, UK
Discussion: *Heloísa Santos*, *Lina Ramos*, *Carolino Monteiro*, *Célia Ventura*

11h00-12h00 – Coffee-break / Poster Viewing

12h00-12h15 – **SPGH Award Lecture**

12h15-12h45 – **SPGH Awards Ceremony**

12h45-13h00 – **Closing Session**