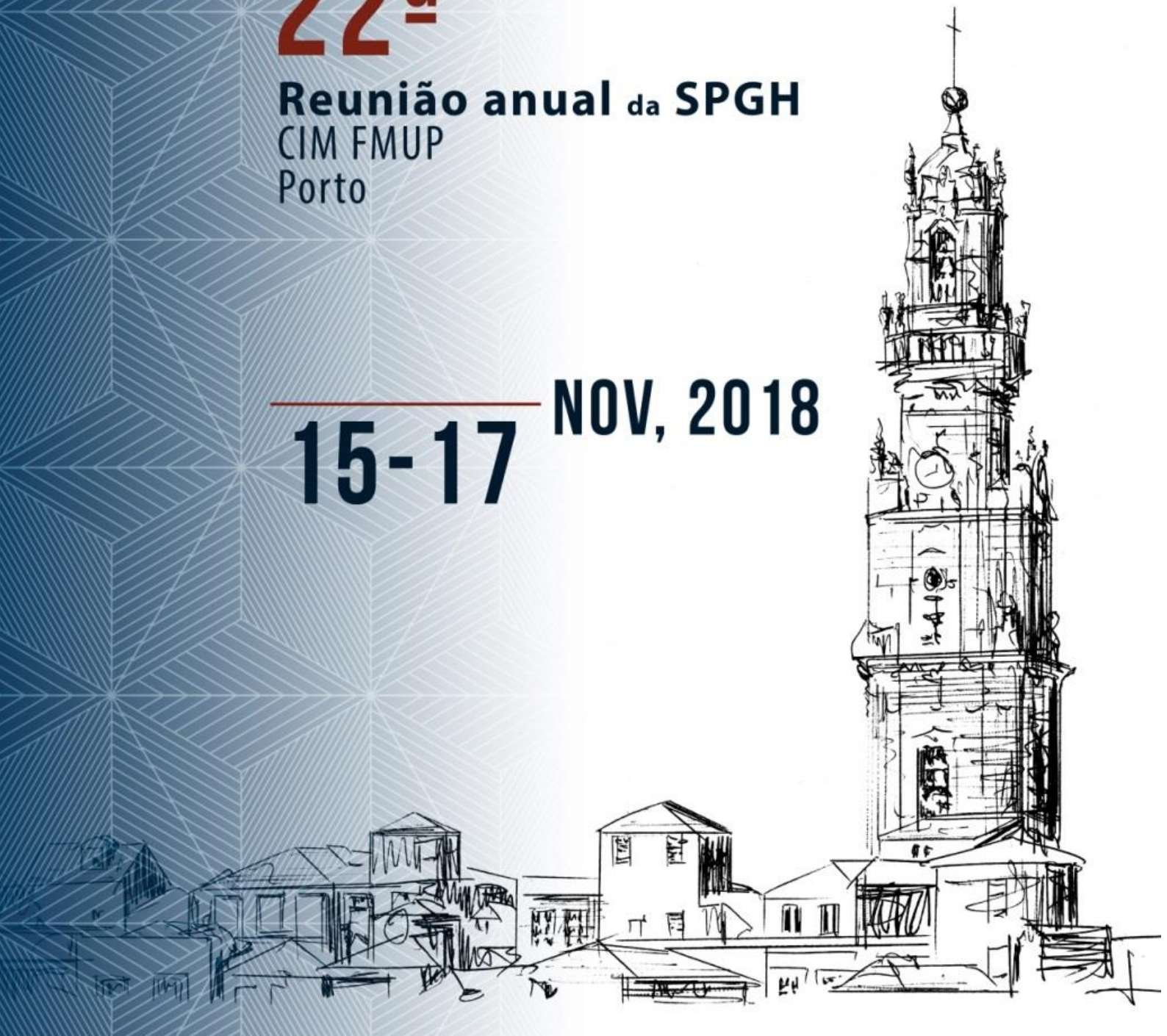
PROGRAMA
PROGRAMME

22ª
Reunião anual da SPGH
CIM FMUP
Porto

15-17 NOV, 2018



THURSDAY 15TH NOVEMBER

09:00h **REGISTRATION**

10:00h **SPGH CLUB MEETINGS** (Concurrent)

MOLECULAR AND CYTOGENETICS CLUB (Room 3)

Pitfalls in genomic analysis
Chairs: Paula Jorge; Cristina Candeias

DYSMORPHOLOGY AND CLINICAL GENETICS CLUB (Room 4)
Chairs: Gabriela Soares; Renata Oliveira; Cláudia Reis

10:00h *Blefarofimose, ptose e outras anomalias palpebrais: revisão do tema e apresentação de casos com diagnóstico*
Ana Rita Soares; **Teresa Saraiva** (S. Genética Médica, CGMJM, CHUP)

10:40h Apresentação e discussão de casos de outros Serviços, subordinados ao tema

12:00h Apresentação e discussão de outros casos, com ou sem diagnóstico

12:30h **SATELLITE MEETING: EUROPEAN REFERENCE NETWORKS IN PORTUGAL** (Room 3)

Panel Discussion: ***What are ERNs and what is in there for me?***
Chairs: Carla Oliveira; Sérgio Sousa

12:30h *What are ERNs and why were they created?*
Carmo Fonseca (Leader, Working Group for Implementation of ERNs in Portugal)

12:45h *Spreading the knowledge at the National level*
Carla Oliveira (National Coordinator GENTURIS ERN; Representative of P.CCC)

12:50h *ERN on bone disorders (ERN BOND) and ERN on congenital malformations and rare intellectual disability (ERN ITHACA)*
Sérgio Sousa (National Coordinator BOND ERN; Representative of CHUC, Coimbra)

13:00h *European Reference Network on genetic tumour risk syndromes (ERN GENTURIS)*
Carla Oliveira (National Coordinator GENTURIS ERN; Representative of P.CCC)

13:05h *The large ERN-collaborative project - Solve-RD*
Carla Oliveira

14:00h **OPENING & WELCOME**
Rosário Santos; João Silva; Rosário Pinto Leite

14:15h **KEYNOTE LECTURE**
Chair: João Silva

The Genetic Landscape of ID
Anita Rauch (Inst. of Medical Genetics, Zurich)

15:00h **EPIGENETICS**
Chairs: Carla Oliveira; Sofia Dória

The role of Imprinting in Cancer
David Monk (Inst. d'Investigació Biomédica, Barcelona)

Imprinting errors in spermatogenic cells of infertile patients
Joana Marques (Dept. Patologia, HSJ, Porto)

Establishing the human epigenome in development and pluripotency
Peter Rugg-Gunn (Babraham Institute, Univ. Cambridge)

16:30h **COFFEE-BREAK / POSTER VIEWING**

16:45h **POSTER HIGHLIGHTS:** Speed talks **(P1-P11)** (AUDITORIUM)

17:30h **SELECTED ORAL PRESENTATIONS (I) _ BASIC RESEARCH (OP1-OP5)**
Chairs: Joana Barbosa de Melo; Susana Fernandes

18:30h **SPGH GENERAL ASSEMBLY**

FRIDAY 16TH NOVEMBER

08:45h **SELECTED ORAL PRESENTATIONS (II) _ CLINICAL RESEARCH (OP6-OP10)**
Chairs: Isabel Marques; Paula Jorge

09:45h ***MEET THE EXPERT*: CLN2, A FORM OF BATTEN DISEASE** (SPONSORED BY BIOMARIN)
Chair: Manuela Santos

An algorithm for CLN2 Diagnosis
Miguel Leão (Dept. Genética Humana, HSJ, Porto)

CLN2 disease- from early diagnosis to early intervention
Eva Wibbeler (Dept. Paediatrics, UMC Hamburg-Eppendorf, Hamburg)

10:30h **COFFEE-BREAK / POSTER VIEWING**

10:45h **POSTER DISCUSSIONS** (e-Posters)

11:15h **IMMUNOTHERAPY AND GENETICS**
Chair: José Carlos Machado

Harnessing the immunotherapy revolution for the treatment of colorectal cancer: neo-antigens and beyond
Noel de Miranda (Leiden Univ. Medical Center, Leiden)

The genetic basis of response to checkpoint inhibitors
Vassiliki Kotoula (AUTH Medical School, Thessaloniki) (SPONSORED BY ASTRAZENECA)

CAR-T for the treatment of T cell malignancies
John F. DiPersio (Alvin J. Siteman Cancer Center / Div. Oncology, Washington Univ. Medical School) (SPONSORED BY FLAD)

13:00h **LUNCH BREAK**

14:00h **NEUROGENETICS AND THERAPY**
Chairs: Jorge Oliveira; Luísa Romão

Towards modifying disease progression and onset in Huntington's Disease and other genetic neurodegenerative disorders: hopes and challenges
Bernhard Landwehrmeyer (Dept. Neurology, Univ. of Ulm)

LRP10: a novel player in late-onset inherited synucleinopathies
Wim Mandemakers (Erusmus Medical Center, Rotterdam)

Fluid Biomarkers in Neurodegenerative Diseases
Luís Maia (Dept. Neurologia, CHUP, Porto)

Advanced therapies in SMA: beyond the clinical trials
Eduardo Tizzano (H. Valle Hebron, Barcelona) (SPONSORED BY BIOGEN)

16.15h **COFFEE-BREAK / POSTER VIEWING**

16:30h **POSTER DISCUSSIONS** (e-Posters)

16:45h **CORPORATE SYMPOSIUM _ AGILENT / SOQUÍMICA**
A clinical case: From the sample to the analysis on the NGS Workflow
Alba Mota; **Ivan Lesende**

17:15h **CORPORATE SYMPOSIUM _ 10X GENOMICS / ILC LDA.**
10x Genomics technology in Human Genetics: from bulk long range sequencing to Copy Number variation in Single Cells
Hannes Arnold

17:45h **KEYNOTE LECTURE**
Chair: Sérgio Sousa

The genetics of cognitive epigenetics
Hans van Bokhoven (Radboud Univ. Medical Center / Donders Inst. for Brain, Cognition & Behaviour, Nijmegen)

20:00h **CONGRESS DINNER**

SATURDAY 17TH NOVEMBER

09:00h **SELECTED ORAL PRESENTATIONS (III) _ CLINICAL CASE REPORTS (OP11-OP18)**
Chairs: Ana Berta Sousa; Lina Ramos

10:00h **BIOETHICS DEBATE** (SPGH BIOETHICS COMMITTEE)
GENOME EDITING – WHAT, WHY AND HOW? THE ETHICAL ISSUES
Chair: Heloísa Santos

CRISPR-Cas 9 – Method and potential applications
André Travessa (S. Genética, Hospital de Santa Maria)

Genome editing - from benefits to ethical limits
Heloísa Santos (SPGH Bioethics Com. / S. Genética, HSM)

Current Portuguese legislation and ethical guidelines
André Pereira (SPGH Bioethics Com. / Fac. Law, U. Coimbra)

11:00h **COFFEE-BREAK / POSTER VIEWING**

11:30h **KEYNOTE LECTURE**
Chair: José Luís Costa

Unlocking the diagnostic potential for circulating DNA
Y.M. Dennis Lo (Fac. of Medicine, The Chinese University of Hong Kong, Shatin, New Territories, Hong Kong SAR, China)
(SPONSORED BY ALFAGENE)

12:15h ***SPGH AWARD* LECTURE**
Chair: Carla Oliveira

A Pentanucleotide ATTTC Repeat Insertion in the Non-coding Region of DAB1, Mapping to SCA37, Causes Spinocerebellar Ataxia
Joana R. Loureiro,***Ana I. Seixas**,* *et al.*
(Am J Hum Genet 2017; 101(1):87–103) *equal contributors

12:30h **SPGH AWARDS CERIMONY**

12:45h **CLOSING SESSION**