SPGH Sociedade Portuguesa de Genética Humana

20ª REUNIÃO ANUAL 2016

Fundação Bissaya Barreto COIMBRA 10-12 de NOVEMBRO 2016

Day 1 Thursday, 10 November

09.30 h - **Registration Opening**

10.00 h **- *Clubs (Parallel Sessions)***

Cytogenetic and Molecular Genetics Club

Medical Genetics and Clinical Dysmorphology Club

14.15 h - ***Opening & Welcome***
Joana Barbosa de Melo, SPGH President
Sérgio Bernardo de Sousa, SPGH Secretary
Cecília Correia, SPGH Treasurer

14.30 h – ***Neurogenetics***
Chairs: Catarina Resende de Oliveira; Lina Ramos

Molecular approaches for Machado-Joseph disease
Luis Almeida, CNC, Faculty of Pharmacy, Coimbra, Portugal

Genomics in neurodegenerative diseases
José Miguel Brás, London, UK

15.30 h**- *Selected Clinical Cases*** *I (see below)*
Chairs: Ana Berta Sousa, Fabiana Ramos

16.00 h - Coffee Break / Poster Viewing

16.25 h – **Corporate Symposium - Illumina**
Furthering clinical testing with genomics solutions from Illumina
Szabolcs Kokeny, PhD., Sr. Sales Product Specialist EMEA, Channel Partners

16.45 h – **Genome structure and phenotype: 16p11.2 rearrangements as an example**
Chair: Joana Barbosa de Melo
Alexandre Reymond, Un. Lausanne, Switzerland

17.15 h - ***Selected Oral Communications I*** *(see below)*
Chairs: Filipa Carvalho, Ana Cristina Sousa

18.45 h - **SPGH Assembly**

Day 2 Friday, 11 November

08.45 h – ***Selected Oral Communications II*** *(see below)*
Chairs: Sofia Dória; Rosario Pinto Leite

10.30 h – **Copy Number Variations (CNVs) can cause disease by changing the 3D-structure of the genome.**
Chair: Isabel Marques Carreira
Stefan Mundlos, Max Plank Institute, Berlin, Germany

11.15 h - Coffee Break / Poster Viewing

11.30 - **Poster discussion with the Scientific Committee**

12.00 h – ***Oncogenetics***
Chairs: Carla Oliveira; José Manuel Nascimento Costa
From Li-Fraumeni syndrome to p53-related cancers
Thierry Frebourg, Rouen University Hospital, Rouen, France

Thyroid cancer: from transformation to (quasi) immortalization
Paula Soares, Ipatimup, Porto, Portugal

13.00 h - Lunch

14.15 h – ***Challenges in the interpretation of sequence variants***
Chairs: Sérgio B Sousa; Susana Fernandes

Implementation of the ACMG Guidelines for Variant Interpretation
Steven Harrison, Harvard University, USA

Towards a quantitative Bayesian pathogenicity and diagnosis framework
Leslie Biesecker, National Human Genome Research Institute, USA

Genetic variants of uncertain clinical significance in hereditary breast cancer: challenges for clinical management
Encarna Gomez, Maastricht, The Netherlands

Classification of genetic variants: lessons and challenges from cardiogenetics
José Carlos Machado, IPATIMUP, Portugal

16.15 h - Coffee Break / Poster Viewing

16.40 h – **Corporate Symposium - Sophia Genetics**
Leveraging the collective knowledge of the largest clinical genomics community to democratize Data-Driven Medicine
Jean-François Vanbellinghen, Subject Matter Expert

17.00 h – ***Prenatal Diagnosis session***
Chairs: Jorge Saraiva; Maria do Céu Almeida
Prenatal: where are we going?
Isabel Marques Carreira, FMUC, Coimbra, Portugal
Fabiana Ramos, HP-CHUC, Coimbra, Portugal

17.45 h– **Public Policy Session "Centros de Referência em Portugal – Estratégia para as Doenças Genéticas"**
Chairs: Jorge Sequeiros; Luísa Romão
Research on Rare Diseases, Reference Centres in Portugal and European Reference Networks: are we following the right strategy to meet our national needs?
Jorge Sequeiros, IBMC, i3S, Porto, Portugal

Reference Centres for rare diseases in Portugal: Will we meet the expectations?
João Lavinha, INSA, Lisboa, Portugal

20.00 h - **Conference Dinner**

Day 3 Saturday, 12 November

08.45 h – ***Selected Clinical Cases*** *II (see below)*
Chairs: Ana Berta Sousa, Jorge Saraiva

09.30 h – ***Bioethics debate:*** **Expanded Carrier Screening – a new tool in Primary Genetic Prevention**
Chairs: Heloísa Santos; Célia Ventura, Francisco Corte Real

Introduction – Heloísa Santos, SPGH, Lisboa, Portugal

Classic carrier screening in Portugal - Haemoglobinopathies – João Lavinha, INSA, Lisboa, Portugal

Responsible implementation of expanded carrier screening – Lidewij Henneman, VU University Medical Center, Amsterdam, The Netherlands

10.30 h – **Sessão Comemorativa do 20º aniversário da SPGH**

10.45 h - Coffee Break / Poster Viewing

11.15 h – **Mosaicism and the Molecular Taxonomy of Human Disease**
Chair: Margarida Reis Lima
Leslie Biesecker, National Human Genome Research Institute, USA

12.15 h – ***SPGH Award Conference***

12.45 h – ***Basic and Clinical Research Awards Ceremony***

12.55 h - ***Closing Session***

Selected oral communications and clinical cases presentations

#	Selected Clinical Cases I, Thursday 10th 15:30	Presenting author
CC1	Case Report of Oculoectodermal Syndrome due to a Mosaic *KRAS* Mutation	André Travessa
CC2	X-Linked Intellectual Disability Syndrome type Nascimento, caused by *UBE2A* mutations: report of two affected brothers and literature review	Sofia Fernandes
CC3	An emerging XLID syndrome affecting females caused by *DDX3X* de novo variants: A case report	Ana Miguel Amaral
CC4	X-linked intellectual disability caused by a novel *PAK3* mutation in a large pedigree	Joana Rosmaninho Salgado
CC5	Comprehensive genomic studies decipher the classical Fragile-X phenotype in a female patient	Paula Jorge

#	Selected Oral Communications I, Thursday 10th 17:15	Presenting author
OC1	Comparison of CRISPR-based methods for modeling loss-of-function in iPS cells	Catarina Seabra
OC2	Non-invasive and viral-mediated silencing of mutant ataxin-3 alleviates motor and neuropathological deficits in a transgenic mouse model of Machado-Joseph disease	Rui Nobre
OC3	Increased frequency of CNVs targeting genes that regulate exposure to toxicants in Autism Spectrum Disorder (ASD): a role for gene-environment interactions	João Pedro Santos
OC4	Array-CGH as a tool in a clinical laboratory set- up: experience in 4000 samples	Susana Ferreira
OC5	Sequence variation between 462 human individuals fine-tunes functional sites of RNA processing	Pedro Ferreira
OC6	Broad multi-gene panel or whole exome sequencing in malformed fetuses reveals five definitive and one likely diagnoses in the first nine cases studied in prenatal setting.	Joaquim Sá

#	Selected Oral Communications II – Friday 10th 08:45	Presenting Author
OC7	Application of whole-genome array CGH un prenatal diagnosis	Alexandra Mascarenhas
OC8	Clinical exome sequencing: the importance of re-analyzing unsolved cases	Fátima Daniela Lopes
OC9	Diagnosis of Mendelian Disorders Using a Comprehensive 4813 Genes Next-Generation Sequencing Panel: Review of 92 Cases	Ana Luísa Carvalho
OC10	The relevance of rare CDH1 non-coding variants in HDGC syndrome: redefining CDH1 cis-regulatory elements	Ana Valente
OC11	Familial intestinal gastric cancer: search for a germline and somatic cause	Joana Carvalho
OC12	Germline variants in homologous recombination (HR)-mediated DNA damage repair genes may contribute to increased colorectal cancer susceptibility in a subgroup of FCCTX families	Ana Magalhães
OC13	Gene expression profiling to predict clinical outcome of oral cavity carcinomas	Ilda Ribeiro

#	Selected Clinical Cases II – Saturday12th 08:45	Presenting Author
CC6	KBG syndrome: the experience of a regional Medical Genetics Unit	Pedro Louro
CC7	Identification of two new candidate genes *OAF* and *PVRL1* for Peters anomaly and ectopia lentis	David Dezdo
CC8	Intermediate Autossomal Recessive Osteopetrosis- Long Term Follow Up on 3 Cases with *CLCN7* mutations	Teresa Carminho
CC9	A new mutation in *ADNP* in a boy with Helsmoortel-Van der Aa syndrome	Mariana Soeiro e Sá
CC10	Beta-propeller protein-associated neurodegeneration (BPAN) in monozygotic twins due to a new mutation *WDR45*	Ana Garabal
CC11	2q31.1 deletion syndrome: report of three patients	Catarina Machado
CC12	A new neuromuscular disorder caused by defects in the activating signal cointegrator 1 complex: the second case with a loss-of-function variant in *ASCC1*	Jorge Oliveira