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; Rosário 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
**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**

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**Selected oral communications and clinical cases presentations**

<table>
<thead>
<tr>
<th>#</th>
<th>Selected Clinical Cases I, Thursday 10th 15:30</th>
<th>Presenting author</th>
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<tbody>
<tr>
<td>CC1</td>
<td>Case Report of Oculoectodermal Syndrome due to a Mosaic KRAS Mutation</td>
<td>André Travessa</td>
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<tr>
<td>CC2</td>
<td>X-Linked Intellectual Disability Syndrome type Nascimento, caused by UBE2A mutations: report of two affected brothers and literature review</td>
<td>Sofia Fernandes</td>
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<tr>
<td>CC3</td>
<td>An emerging XLID syndrome affecting females caused by DDX3X de novo variants: A case report</td>
<td>Ana Miguel Amaral</td>
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<tr>
<td>CC4</td>
<td>X-linked intellectual disability caused by a novel PAK3 mutation in a large pedigree</td>
<td>Joana Rosmaninho Salgado</td>
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<tr>
<td>CC5</td>
<td>Comprehensive genomic studies decipher the classical Fragile-X phenotype in a female patient</td>
<td>Paula Jorge</td>
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<tr>
<td>#</td>
<td>Selected Oral Communications I, Thursday 10th 17:15</td>
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<tr>
<td>OC1</td>
<td>Comparison of CRISPR-based methods for modeling loss-of-function in iPS cells</td>
<td>Catarina Seabra</td>
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<tr>
<td>OC2</td>
<td>Non-invasive and viral-mediated silencing of mutant ataxin-3 alleviates motor and neuropathological deficits in a transgenic mouse model of Machado-Joseph disease</td>
<td>Rui Nobre</td>
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<tr>
<td>OC3</td>
<td>Increased frequency of CNVs targeting genes that regulate exposure to toxicants in Autism Spectrum Disorder (ASD): a role for gene-environment interactions</td>
<td>João Pedro Santos</td>
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<tr>
<td>OC4</td>
<td>Array-CGH as a tool in a clinical laboratory set-up: experience in 4000 samples</td>
<td>Susana Ferreira</td>
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<td>OC5</td>
<td>Sequence variation between 462 human individuals fine-tunes functional sites of RNA processing</td>
<td>Pedro Ferreira</td>
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<tr>
<td>OC6</td>
<td>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.</td>
<td>Joaquim Sá</td>
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<th>#</th>
<th>Selected Oral Communications II – Friday 10th 08:45</th>
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<tbody>
<tr>
<td>OC7</td>
<td>Application of whole-genome array CGH un prenatal diagnosis</td>
<td>Alexandra Mascarenhas</td>
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<tr>
<td>OC8</td>
<td>Clinical exome sequencing: the importance of re-analyzing unsolved cases</td>
<td>Fátima Daniela Lopes</td>
</tr>
<tr>
<td>OC9</td>
<td>Diagnosis of Mendelian Disorders Using a Comprehensive 4813 Genes Next-Generation Sequencing Panel: Review of 92 Cases</td>
<td>Ana Luisa Carvalho</td>
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<tr>
<td>OC10</td>
<td>The relevance of rare CDH1 non-coding variants in HDGC syndrome: redefining CDH1 cis-regulatory elements</td>
<td>Ana Valente</td>
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<tr>
<td>OC11</td>
<td>Familial intestinal gastric cancer: search for a germline and somatic cause</td>
<td>Joana Carvalho</td>
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<td>OC12</td>
<td>Germline variants in homologous recombination (HR)-mediated DNA damage repair genes may contribute to increased colorectal cancer susceptibility in a subgroup of FCCTX families</td>
<td>Ana Magalhães</td>
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<td>OC13</td>
<td>Gene expression profiling to predict clinical outcome of oral cavity carcinomas</td>
<td>Ilda Ribeiro</td>
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<tr>
<td>CC6</td>
<td>KBG syndrome: the experience of a regional Medical Genetics Unit</td>
<td>Pedro Louro</td>
</tr>
<tr>
<td>CC7</td>
<td>Identification of two new candidate genes OAF and PVAL1 for Peters anomaly and ectopia lentis</td>
<td>David Dezdo</td>
</tr>
<tr>
<td>CC8</td>
<td>Intermediate Autosomal Recessive Osteopetrosis- Long Term Follow Up on 3 Cases with CLCN7 mutations</td>
<td>Teresa Carminho</td>
</tr>
<tr>
<td>CC9</td>
<td>A new mutation in ADNP in a boy with Helsmoortel-Van der Aa syndrome</td>
<td>Mariana Soeiro e Sá</td>
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<tr>
<td>CC10</td>
<td>Beta-propeller protein-associated neurodegeneration (BPAN) in monozygotic twins due to a new mutation WDR45</td>
<td>Ana Garabal</td>
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<tr>
<td>CC11</td>
<td>2q31.1 deletion syndrome: report of three patients</td>
<td>Catarina Machado</td>
</tr>
<tr>
<td>CC12</td>
<td>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</td>
<td>Jorge Oliveira</td>
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