10h00  **WELCOME**  
**Chairs:** João Gonçalves, Célia Ventura, Rosário Pinto Leite

10h10 - 10h40  **KEYNOTE LECTURE 1 “A DIAGNOSIS FOR ALL RARE GENETIC DISEASES”**  
(virtual communication)  
Kym Boycott (CHEO Research Institute, University of Ottawa, Ottawa, Canada)  
**Chair:** Carla Oliveira

10h40 - 11h00  **Coffee-break / ePoster viewing / Concurrent Supporting Companies’ presentations**

11h00 - 11h50  **SELECTED ORAL PRESENTATIONS - SESSION I: BASIC RESEARCH**  
(virtual communications with live questions and answers)  
**Chairs:** Joana Melo and Susana Fernandes

**OP1**  
**GENE EDITED IPS-C-DERIVED CARDIOMYOCYTES REVEAL RNA SPLICING DEFECTS IN HYPERTROPHIC CARDIOMYOPATHY**  
Marta Furtado  
Instituto de Medicina Molecular João Lobo Antunes, Faculdade de Medicina, Universidade de Lisboa  
Lisboa, Portugal

**OP2**  
**PERMANENT INACTIVATION OF THE ATXN3 GENE: A POSSIBLE THERAPEUTIC APPROACH FOR MACHADO-JOSEPH DISEASE**  
Sara M. Lopes  
Center for Neuroscience and Cell Biology (CNC), University of Coimbra, Portugal;  
Institute for Interdisciplinary Research (IIIUC), University of Coimbra, Coimbra, Portugal

**OP3**  
**THE TUMOR SUPPRESSOR P53 ACQUIRES ONCOGENIC FUNCTIONS DUE TO A TRANSLATIONAL SWITCH DURING INTEGRATED STRESS RESPONSE**  
Rafaela Lacerda  
MaRCU — Molecular and RNA Cancer Unit; Departamento de Genética Humana,  
Instituto Nacional de Saúde Dr. Ricardo Jorge, Lisboa, Portugal;  
BioISI — BioSystems & Integrative Sciences, Faculdade de Ciências, Universidade de Lisboa, Lisboa, Portugal

11h55 - 12h40  **SELECTED ORAL PRESENTATIONS - SESSION II: CLINICAL RESEARCH**  
(virtual communications with live questions and answers)  
**Chairs:** Sérgio Sousa and Sofia Dória

**OP4**  
**GENOTYPE-PHENOTYPE ASSOCIATIONS PROVIDE A RATIONAL TO IDENTIFY POTENTIALLY ACTIONABLE VUS**  
José Peláez  
IPATIMUP i3, Porto, Portugal

**OP5**  
**EFFECT OF FMR1 ALLELIC COMPLEXITY COMBINATIONS IN X-CHROMOSOME INACTIVATION SKEWING: TESTS ON COHORTS OF INFERTILE FEMALES AND OOCYTE DONORS**  
Bárbara Rodrigues  
Molecular Genetics Unit, Centro de Genética Médica Jacinto Magalhães (CGM),  
Centro Hospitalar Universitário do Porto (CHUP); Unit for Multidisciplinary Research in Biomedicine (UMIB),  
Institute of Biomedical Sciences Abel Salazar, (ICBAS), UP, Porto, Portugal

**OP6**  
**CELL-FREE DNA: A TOOL FOR THE DIAGNOSIS AND FOLLOW-UP OF ORAL CANCER?**  
Ivana Martins  
Cytogenetics and Genomics Laboratory, Faculty of Medicine, University of Coimbra, Coimbra, Portugal
SELECTED ORAL PRESENTATIONS - SESSION III: CLINICAL CASES
(virtual communications with live questions and answers)

Chairs: Ana Berta Sousa and Lina Ramos

OP7 A CASE OF MOWAT-WILSON SYNDROME DUE TO A MISSENSE VARIANT - CASE REPORT AND REVIEW OF THE LITERATURE
Susana L. Ferreira
Serviço de Genética Médica, Centro Hospitalar Universitário de Lisboa Central, Lisboa, Portugal

OP8 CUBILIN VARIANTS IN PATIENTS WITH CHRONIC KIDNEY DISEASE INCLUDING END-STAGE
Rita Quental
Serviço de Genética Médica, Centro Hospitalar Universitário de São João, Porto, Portugal

OP9 TBCD-RELATED ENCEPHALOPATHY: A PATIENT WITH A SEVERE NEONATAL PRESENTATION
Marta P. Soares
Serviço de Genética, Hospital de Santa Maria, Centro Hospitalar Universitário Lisboa Norte, Centro Académico de Medicina de Lisboa, Lisboa, Portugal

Lunch-break / ePoster viewing / Concurrent Supporting Companies’ presentations

14h00 - 14h30 SPONSOR’S PRESENTATIONS

14h00 - 14h15 KAPA TARGET ENRICHMENT PORTFOLIO - BETTER BY DESIGN
Markos Mihalatos

14h15 - 14h30 SCREENING OF NEW DISEASE-CAUSING GENES IMPROVES THE MOLECULAR DIAGNOSTIC YIELDS OF EXOME SEQUENCING-NEGATIVE CASES WITH INTELLECTUAL DISABILITY
Joana Salgado (CHUC)

(virtual communications with live questions and answers)

Chair: Célia Ventura

BE-1: “THE NEW THERAPIES AND THE DIFFICULT CHALLENGE OF EQUITY AND BENEFICENCE IN HEALTHCARE. THE ROLE OF SOCIETY”
Heloísa Santos (President of the Bioethics Commission of SPGH)

BE-2: “EQUITY, JUSTICE AND BENEFICENCE IN ACCESS TO PHARMACEUTICAL DRUGS”
António Faria Vaz (INFARMED, Lisbon, Portugal)

BE-3: “AND THE SOCIETY?”
Joaquim Brites (APN - Associação Portuguesa de Neuromusculares)

Conclusions and Recommendations
Heloísa Santos (President of the Bioethics Commission of SPGH)
Carolino Monteiro (Faculty of Pharmacy, University of Lisbon, Portugal)

General Discussion (live questions and answers)
15h30 - 16h00  **KEYNOTE LECTURE 2: “INBORN ERRORS OF IMMUNITY TO SARS-COV-2”**  
(virtual communication)  
Jean-Laurent Casanova (The Rockefeller University, New York, USA)  
**Chair:** Isabel Carreira

16h00 - 16h30  **Coffee-break / ePoster viewing / Concurrent Supporting Companies’ presentations**

16h30 - 17h00  **SPGH AWARD LECTURE**  
(virtual communication)  
**Speaker to be announced**  
**Chairs:** Carla Oliveira and Jorge Pinto Basto

17h00 - 17h20  **SPGH AWARDS CEREMONY**  
(live session)  
**Chairs:** João Gonçalves and Carla Oliveira

17h20 - 17h30  **CLOSING SESSION**  
(live session)  
**Chairs:** João Gonçalves, Célia Ventura, Rosário Pinto Leite  
**Address by the Future President**

17h45 - 20h00  **SPGH GENERAL ASSEMBLY**  
**Chairs:** João Gonçalves, Célia Ventura, Rosário Pinto Leite