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24TH ANNUAL MEETING SOCIEDADE PORTUGUESA DE GENÉTICA HUMANA

NOV. 20TH 2020

(VIRTUAL MEETING)

PROGRAMME



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Instituto Nacional de Saúde
Doutor Ricardo Jorge

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FACULDADE DE MEDICINA
UNIVERSIDADE DE
COIMBRA



INSTITUTO DE INVESTIGAÇÃO
E INOVAÇÃO EM SAÚDE
UNIVERSIDADE DO PORTO



ORDEM DOS
BIÓLOGOS





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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 IPSC-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



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12h45 - 13h15 **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 TB CD-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*

13h15 - 14h30 **Lunch-break / ePoster viewing / Concurrent Supporting Companies' presentations**

14h00 - 14h30 **SPONSOR'S PRESENTATIONS**

14h00 - 14h15 **KAPA TARGET ENRICHMENT PORTFOLIO
- BETTER BY DESIGN**

Markos Mihalatos

Sponsor:



14h15 - 14h30 **SCREENING OF NEW DISEASE-CAUSING GENES
IMPROVES THE MOLECULAR DIAGNOSTIC YIELDS
OF EXOME SEQUENCING-NEGATIVE CASES WITH
INTELLECTUAL DISABILITY**

Joana Salgado (CHUC)

Sponsor:



14h30 - 15h30 **BIOETHICS DEBATE (BE): "SENSE AND SENSIBILITY – THE PRINCIPLE OF EQUITY
AND THE DIFFICULT BALANCE OF THE NEW TARGETED THERAPIES FOR GENETIC DISEASES"**

(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)



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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



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EXECUTIVE SECRETARIAT

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