

5TH GENOME PT SYMPOSIUM

ADVANCING GENOMICS
FOR A HEALTHIER FUTURE

 **1 JULY 2026**

 **FACULDADE DE CIÊNCIAS
UNIVERSIDADE DE LISBOA**
LISBON, PORTUGAL

FINAL PROGRAMME

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HOSTING AND
ORGANISATIONAL SUPPORT

MORNING SESSION

FROM GENOMES TO PRECISION HEALTH CARE

- 09:00 • **REGISTRATION**
- 09:30 • **WELCOME**
GenomePT & CIÊNCIAS ULisboa representatives
- 09:45 • **GENOMEPT 2016-2026**
Building genomics capacity in Portugal
Margarida Gama Carvalho, GenomePT Coordinator
- 10:00 • **i3S-HEALTH: Teaming to Drive the Future of Genomic Medicine**
Cláudio Sunkel, Director, I3S
- 10:20 • **PT_MedGen: National Strategy for Genomic Medicine**
Astrid Vicente, PT_MedGen Coordinator, INSA
- 10:40 • **ROUND TABLE**
Building Genomic Medicine in Portugal: Strategies, Infrastructures and Implementation
Moderated by Manuel Santos (Director, MIA, UCoimbra)
Panel: Astrid Vicente (INSA), Cláudio Sunkel (i3S), Jorge Oliveira (BioData), Sérgio Sousa (ULS Coimbra), Margarida Gama Carvalho (GenomePT)
- 11:15 • **COFFEE-BREAK AND POSTERS**
- 11:45 • **Genetic test clinical utility and variant (re)classification: a long-read story and beyond**
Jorge Oliveira, CGPP - IBMC
- 12:05 • **Learning hub in rare diseases and the Preventable Project**
Carla Oliveira, I3S

SELECTED ORAL COMMUNICATIONS

- 12:25 • **Transcriptional and Chromatin Determinants of Human Naïve Regulatory CD4 T Cell Identity**
Alexandre Raposo, Gulbenkian Institute for Molecular Medicine
- 12:35 • **Medieval Iasi Through the Lens of Ancient DNA: Population Structure, Ancestry and Cemetery-Level Variation**
João Brochado de Almeida, Universidade do Minho
- 12:45 • **Detection of the BRCA2 c.156_157insAlu germline variant from FFPE tumor samples DNA using short-read targeted NGS**
Manuela Pinheiro, IPO-Porto
- 12:55 • **LUNCH AND POSTER SESSION**

AFTERNOON SESSION

FROM GENOMES TO SOCIETAL IMPACT

- 14:30 • **EASIGEN-DS Consortium – Design Study for a Research Infrastructure on Advanced Genomics Technologies**
Ivo Gut, Director, Centro Nacional de Análisis Genómico (CNAG), Spain
- 15:00 • **Metabolic innovation through horizontal gene transfer in yeasts**
Paula Gonçalves
UCIBIO, FCT-UNL

TECHNOLOGY SPOTLIGHTS

- 15:20 • **Sequencing reimaged: introducing the Illumina TruPath Genome**
Stef Kupers, Illumina
- 15:40 • **Beyond the Blind Spots: What Long-Read Sequencing is changing in Human Genomics**
Ximo García, PacBio

16:00 • COFFEE-BREAK AND POSTERS

- 16:30 • **Past Genomes, Future Conservation: Museomics to the rescue**
Ricardo Lopes, Ce3C, CIÊNCIAS ULisboa

TECHNOLOGY SPOTLIGHTS

- 16:50 • **New quality standard of sequencing with Aviti**
Agnieszka Ciesielska, Element Bioscience
- 17:10 • **Rethinking WGS with MGI: Why Quality, Speed, and Cost No Longer Require Trade-offs**
Bernard Okere, MGI
- 17:30 • **Setting new boundaries for single-cell and spatial multiomics analysis with 10X Genomics**
Ana Borges, Bonsailab

SELECTED ORAL COMMUNICATIONS

- 17:45 • **A genome language model for mapping DNA replication origins**
Francisco Berke meier, University of Cambridge
- 17:55 • **Transcriptomic signatures differentiating the successes and limitations of regeneration in a marine annelid**
Inês Moutinho Cabral, UCIBIO, FCT-UNL
- 18:05 • **Single nucleus transcriptome atlas of the honey bee gut**
Waldan Kwong, Gulbenkian Institute for Molecular Medicine
- 18:15 • **CLOSING**

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