

Symposium partners



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# Genome<sup>PT</sup> SYMPOSIUM

2025  
May 30<sup>th</sup>

4<sup>th</sup> Symposium of the National Research  
Infrastructure for Genome Sequencing and Analysis



Programme

9h00 Registration

9h30 Welcome (Margarida Gama Carvalho e Nuno Ferrand)

9h40 GenomePT: Current Landscape and Future Directions (Margarida Gama Carvalho)

10h00 European Efforts for Biodiversity Genomics Research (Robert Waterhouse, Chairman of ERGA, Swiss Institute of Bioinformatics - ONLINE)

10h30 Biogenome Portugal: the emerging Portuguese node of the European Reference Genome Atlas (ERGA) (José Melo Ferreira, CIBIO-BIOPOLIS/Ricardo J. Lopes, FCUL, CE3C e MUHNAC)

10h50 *Coffee-break*

11h05 ORAL COMMUNICATIONS 1

11h55 Combined analysis of chromatin accessibility, promoter interactions and whole genome sequencing solved the missing heritability in gastric cancer (C. São José, M. Ferreira, L. Cordova, A. Pedro, J. Senz, J. Garcia-Pelaez, S. Lobo, I. Gullo, A. André, P. Kaurah, F. Puntieri, J. Glaser, D. Huntsman, S. Mundlos, K. Schrader, C. Oliveira)

12h15 Genomic-Derived Data Predicts Functional Impairment in COPD (Rui Marçalo, Guilherme Rodrigues, Miguel Pinheiro, Sonya Neto, Sofia L. Marques, Paula Simão, Vitória Martins, Lília Andrade, Maria Aurora Mendes, Manuel A. S. Santos, Vera Afreixo, Alda Marques, Gabriela R. Moura)

12h35 *Lunch and Poster session*

14h00 Unlocking the Blue Hidden Majority: Transforming Biomonitoring, Food Safety, and Planetary Health in Coastal Areas (Bernardo Duarte, Assistant Professor Faculty of Sciences of the University of Lisbon | BIOTOX - Biomonitoring and Ecotoxicology Laboratory MARE - Marine and Environmental Sciences Centre & ARNET - Aquatic Research Network (LA))

14h20 The possibilities of long-read sequencing for genetic diagnostics (Christian Gilissen, Professor of Genome Bioinformatics (Human Genetics), Sint Radboud University Medical Centre)

14h40 Recent Progress in Reference Genome Assembly Pipelines (Tyler Alioto, Chair of the Sequencing and Assembly Committee, ERGA - ONLINE)

15h00 Experience of a medical genetics laboratory in NGS methodologies and the clinical importance of having a high-throughput NGS workflow accredited to the NP EN ISO 15189 standard (Rita Cerqueira, CGC Genetics Laboratory Director, Unilabs Portugal)

15h20 *Coffee-break*

15h35 The Aviti24™ System: Revolutionizing Multiomics – An Integrated Solution for Genomics, Transcriptomics, and Proteomics (Sebastián Celis-Field Application Scientist, Element Biosciences)

15h55 Advancing NGS in complex and Vestigial Samples: Towards Better Diagnostics and Surveillance (Ricardo Dias, Coordinator | iXLab – Initiative for National Biological Resilience Co-coordinator | Genomic Facility Faculty of Sciences – University Lisbon)

16h15 HiFi Sequencing in Population Genomics: A Global Perspective on Leading Projects and Innovations (Sukhvinder Nicklen - Human Genetics Market Development Manager EMEA at PacBio)

16h35 ORAL COMMUNICATIONS 2

17h10 Closing session

17:15 Nodes Coordinator's Meeting – private