



Societat Catalana
de **BIOLOGIA**



BIOINFORMATICS
BARCELONA

XIII Jornada de Bioinformàtica i Genòmica

Organitzada per:

Secció de Bioinformàtica i Genòmica de la SCB
Associació Bioinformatics Barcelona – BIB

Patrocinada per:



Facultat
Biociències
UAB

UAB
Universitat Autònoma
de Barcelona

PROGRAMA

Facultat de Biociències
Universitat Autònoma de Barcelona

Edifici C, Campus UAB, Bellaterra

18 i 19 de desembre de 2025

COMITÈ ORGANITZADOR:

Jaime Martínez Urtaza (UAB)
Marta Puig (UAB)
Mario Cáceres (ICREA, HMRIB)
Roderic Guigó (CRG-UPF)

SUPORT:

Mariàngels Gallego (SCB)
Paqui Lorite (SCB)
Rosa Bover (BIB)

PROGRAM (December 18th)

9:30 – 9:45 Welcome and opening of the symposium

SESSION 1

9:45 – 10:30 **Invited lecture: Tobias Marschall (Heinrich Heine University).** TBD.

10:30 – 10:45 **Adrià Mompert (HMRIB).** Accurate imputation of inversions in human genomes using different algorithms and data sources.

10:45 – 11:00 **José Marcos Moreno-Cabrera (IDIBELL).** Detection of germline CNVs from gene panel data: benchmarking the state of the art.

11:00 – 11:15 **Montserrat Palazón-Balmaseda (BSC).** Machine-learning powered hash-indexes for high-throughput genome read mapping.

11:15 – 11:45 Coffee break

SESSION 2

11:45 – 12:00 **Max Ticó (UB).** Pyranges v1: a Python framework for ultrafast sequence interval operations.

12:00 – 12:15 **Jaume Reig Palou (UB).** Population prevalence of postzygotic variants at preclinical frequency in the *UBA1* gene.

12:15 – 12:30 **Caterina Fuses (UB).** Context-dependent genetic modifiers of Huntington's disease revealed through multimodal machine learning.

12:30 – 12:45 **Xavier Farré (IGTP).** Sex-specific VEGFA overexpression linked to Long COVID persistence.

12:45 – 13:00 **Xavier Hernández-Alias (MPIB).** Deep learning modelling of mRNA translation adaptation to nutrient shifts reveals novel regulatory elements.

13:00 – 13:15 **Maximilian Stammnitz (CRG).** The genetic architecture of allosteric plant hormone receptors.

13:15 – 13:30 **Sara Martínez de Cripán (EURECAT).** A method for molecular substructure-based in-source fragment annotation.

13:30 – 14:30 Lunch

POSTER SESSION

14:30 – 15:30 Poster viewing with authors

SESSION 3

15:30 – 16:15 **Invited Lecture: Marcos Díaz-Gay (Centro Nacional de Investigaciones Oncológicas).** Uncovering the global genomic landscape of colorectal and lung cancers through mutational epidemiology.

16:15 – 16:30 **Joan Antoni Martorell (HMRIB).** Stop-loss mutations in cancer samples are over-represented in oncogenes and tumor suppressor genes.

16:30 – 16:45 **Guillermo Prol-Castelo (BSC).** Integrating generative AI and symbolic reasoning for kidney cancer progression forecasting.

16:45 – 17:00 **Gerard Martínez-Cebrián (IJC).** Multi-omics integrative analysis identifies synergistic mechanisms between GATA1s and STAG2 mutations in myeloid leukemia of Down syndrome.

17:00 – 17:30 Coffee break

SESSION 4

17:30 – 17:45 **Laura González-Rodelas (IBB-UAB).** Twists in the shell – Genome reshuffling and 3D chromatin architecture in turtles.

17:45 – 18:00 **Ainhoa López (UB).** High-quality chromosome-level genome assemblies and annotations of two Mediterranean wrasse fish.

18:00 – 18:15 **Marc Pulido (CRAG).** Uncovering retrotransposon-driven genome structure evolution in the moss *Physcomitrium patens*.

18:15 – 18:30 **Tomás Montserrat-Ayuso (CNAG).** HERVarium: A unified resource for protein-domain and regulatory annotation of human endogenous retroviruses.

18:30 – 18:45 **Sara Peregrina (BSC).** Oral microbiome signatures of healthy aging: Insights from dementia, physical exercise and extreme longevity.

18:45 – 19:00 **Julia Ortís Sunyer (University of Luxembourg).** PathoFact 2.0: An integrative pipeline for predicting antimicrobial resistance genes, virulence factors, toxins and biosynthetic gene clusters in metagenomes.

19:00 – 20:00 Cocktail and poster viewing

PROGRAM (December 19th)

SESSION 5

- 9:30 – 10:15 **Invited lecture: Maria Colomé-Tatché (Hospital del Mar Research Institute, Ludwig-Maximilians-Universität München, Helmholtz Zentrum München).** Studying single-cell heterogeneity with epigenomic data.
- 10:15 – 10:30 **Albert Alegret-Garcia (ISGlobal).** Empowering rare-disease diagnostics through epigenomics: A novel Mowat-Wilson episignature.
- 10:30 – 10:45 **Vasilis Ntasis (CRG).** Trajectory-based causality analysis reveals bidirectional regulation between histone PTMs and gene expression.
- 10:45 – 11:00 **Akshay Jaya Ganesh (UB).** Single-cell transcriptomics identifies necroptotic MEG3+ excitatory neurons in an iPSC model of Alzheimer's Disease.
- 11:00 – 11:15 **Flavio Palmieri (UB).** Spatial transcriptomic insights into adipocyte hypertrophy during childhood obesity.
- 11:15 – 11:45 Coffee Break and poster viewing

SESSION 6

- 11:45 – 12:00 **Jordi Manuel Cabrera-Gumbau (UAB).** Popkaryote, a novel tool to infer population genomics statistics from prokaryotic organisms.
- 12:00 – 12:15 **Carlos Sarabia (IBB-UAB).** PSMCmast: A novel approach to correct for false negative rates in low-coverage demographic inference.
- 12:15 – 12:30 **Maria Rosselló (UB).** Sex-specific turbanate eye in mayflies reveals a *dsx* duplication underlying its development.
- 12:30 – 12:45 **Miquel Àngel Schikora-Tamarit (BSC).** Unraveling the signs of genomic adaptation to a Neolithic lifestyle.
- 12:45 – 13:30 **Invited Lecture: Sandra Álvarez-Carretero (University College London).** Inferring evolutionary timelines with phylogenomic data: a Bayesian sequential-subtree approach.
- 13:30 – 14:00 Best oral communication and poster awards

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