



LRUA 24

LONG-READ SEQUENCING UPPSALA



PROGRAM

LRUA24, Oct 21-23 2024

lrua2024.se

Uppsala University Main Building, Uppsala, Sweden

MONDAY, OCTOBER 21

9:30 - 10:30 REGISTRATION & COFFEE

LONG-READ SEQUENCING UPDATES

CHAIR: Olga Vinnere Pettersson

- 10:30 - 10:50 Welcome & Introduction to LRUA24
Adam Ameer & Olga Vinnere Pettersson, SciLifeLab Genomics, Sweden
- 10:50 - 11:10 Updates from Oxford Nanopore Technologies
Rosemary Sinclair Dokos, Oxford Nanopore Technologies, UK
- 11:10 - 11:30 Elevate your research with PacBio HiFi sequencing
Somar Al-Walai, PacBio, Sweden

BIODIVERSITY

- 11:30 - 12:00 Tree of life biodiversity sequencing - Balancing quality and quantity
Kerstin Howe, Wellcome Sanger Institute, UK

12:00 - 13:30 LUNCH & POSTERS

BIODIVERSITY continued

CHAIR: Lisa Klasson

- 13:30 - 13:50 Characterisation of particularly complex genomic regions under strong natural selection in Atlantic herring
Mats Pettersson, Uppsala University, Sweden
- 13:50 - 14:10 Atlas of telomeric repeat diversity in *Arabidopsis thaliana*
Yueqi Tao, Max Planck Institute for Biology Tübingen, Germany
- 14:10 - 14:30 High-throughput recovery of microbial genomes from complex soil communities with deep, long-read Nanopore sequencing
Mantas Sereika, Aalborg University, Denmark
- 14:30 - 14:50 Assembling an atlas of European reference genomes takes a whole community
Robert Waterhouse, SIB Swiss Institute of Bioinformatics, Switzerland
- 14:50 - 15:00 Flash talks, round I
Gregor Diensthuber, Vendra Mangkusaputra, Charlotte Van Dijk, Nadja Nolte

15:00 - 15:40 COFFEE & POSTERS

WHAT'S NEXT IN LONG-READ SEQUENCING?

CHAIR: Carl-Johan Rubin

- 15:40 - 16:10 The complete sequence and comparative analysis of ape sex chromosomes
Kateryna Makova, Penn State University, USA
- 16:10 - 16:30 TLDR: Nanopore adaptive sampling strategies for research and the clinic
Matt Loose, Nottingham University, UK
- 16:30 - 16:50 Mapping parent of origin methylation by long-read sequencing reveals novel imprinting and insight into human disease
Elin Grundberg, Children's Mercy Kansas City, USA
- 16:50 - 17:10 MicroST: a scalable platform for efficient spatial long-read sequencing
David McKellar, New York Genome Center, USA

18:30 - late Conference dinner at Norrlands Nation
Västra Ågatan 14, Uppsala

**TUESDAY, OCTOBER 22****TECHNOLOGY & APPLICATIONS**

- 09:00 - 09:30 Decoding the central dogma with single molecules **CHAIR: Susan Kloet**
Winston Timp, Johns Hopkins University, USA
- 09:30 - 09:50 Mapping the adaptive immune repertoire using spatial transcriptomics and long-read sequencing
Kim Thrane, KTH Royal Institute of Technology, Sweden
- 09:50 - 10:10 From fragmented to full-length: A new era in human mitochondrial genome sequencing
Marta Gut, CNAG-CRG & The Barcelona Institute of Science and Technology, Spain
- 10:10 - 10:20 Silver sponsor I
Diagenode
- 10:20 - 10:50 **COFFEE & POSTERS**

HUMAN/MEDICAL SEQUENCING

- 10:50 - 11:20 High resolution assessment of rare genetic disease **CHAIR: Lars Feuk**
Tomi Pastinen, Children's Mercy Kansas City, USA
- 11:20 - 11:40 HiFi long-read genomes for difficult-to-detect clinically relevant variants
Lisenka Vissers, Radboud University, The Netherlands
- 11:40 - 12:00 Effects of SF3B1 mutations in CLL and MDS patients uncovered by long-read transcriptome sequencing
Ralf Herwig, Max-Planck-Institute for Molecular Genetics, Germany
- 12:00 - 12:10 Flash talks, round 2
Anika John, Netanya Keil, Marlene Ek, Emmy Borgmästars

12:10 - 13:30 LUNCH & POSTERS**HUMAN/MEDICAL SEQUENCING continued**

- 13:30 - 13:50 Ultra-fast deep-learned CNS tumor classification during surgery **CHAIR: Ulf Gyllensten**
Jeroen de Ridder, UMC Utrecht, The Netherlands
- 13:50 - 14:10 Short tandem repeat variation in frontotemporal dementia
Wouter De Coster, VIB & University of Antwerp, Belgium
- 14:10 - 14:30 Towards routine long-read sequencing for rare disease, a national pilot study on chromosomal rearrangements
Anna Lindstrand, Karolinska Institutet & Karolinska University Hospital, Sweden
- 14:30 - 14:50 Increasing diagnostic yield by combined long-read RNAseq and WGS in unsolved genetic disorders
Tjakko Van Ham, Erasmus MC, The Netherlands
- 14:50 - 15:00 Enhancing long-read sequencing: Sample preparation strategies for microbiomics and genomics
Henriette Kümmel, Zymo Research Europe GmbH, Germany

15:00 - 15:40 COFFEE & POSTERS**RNA & SINGLE CELL SEQUENCING****CHAIR: Wilfried Haerty**

- 15:40 - 16:10 Using lrrNA-seq in multi-sample experiments: design and bias considerations
Ana Conesa, CSIC Valencia, Spain
- 16:10 - 16:30 Towards a comprehensive single-cell picture of RNA isoforms in mouse and human brain and their diseases – or – single-cell isoforms in time and space
Hagen Tilgner, Weill Cornell Medicine, US
- 16:30 - 16:50 In-depth transcriptome profiling of the motor cortex in ALS/FTLD using an integrative long-read RNA sequencing approach
Isabell Cordts, Mayo Clinic, USA & Technical University of Munich, Germany
- 16:50 - 17:10 Uncovering the intact extracellular transcriptome of liquid biopsies and their RNA-carrying macromolecules
Jasper Verwilt, VIB & University of Antwerp, Belgium
- 17:10 - 17:20 Prize ceremony - Flash talks
Ida Höjjer, SciLifeLab Genomics & Clinical Genomics Uppsala, Sweden

**WEDNESDAY, OCTOBER 23****OXFORD NANOPORE TECHNOLOGIES WORKSHOP**

- 09:00 - 09:15 Introduction
Jakob Ørtvig, Oxford Nanopore Technologies, Denmark
- 09:15 - 09:40 Novel approach to molecular pathology with nanopore sequencing
Skarphéðinn Halldórsson, Oslo University Hospital, Norway

PRODUCT SHOWCASE - Ease of nanopore sequencing

- 09:40 - 09:50 On stage PromethION sequencing set up
Andreas Venizelos, Oxford Nanopore Technologies, Denmark
Christos Coucoravas, Oxford Nanopore Technologies, Sweden
- 09:50 - 10:10 EPI2ME bioinformatics workflow demonstration
Andreas Venizelos, Oxford Nanopore Technologies, Denmark
Christos Coucoravas, Oxford Nanopore Technologies, Sweden
- 10:10 - 10:15 Concluding remarks of the workshop

10:15 - 10:45 COFFEE & POSTERS**PACBIO WORKSHOP: From now to next – The future of genome sequencing is closer than you think**

- 10:45 - 10:50 Introduction
Mike Erbele, PacBio, USA
- 10:50 - 11:05 Building and training tools for the complete genome
Mike Erbele, PacBio, USA
- 11:05 - 11:25 PacBio HiFi long-read genomes offer better exomes by unlocking retinal disease variants missed by short-read sequencing
Christian Betz, Bioscientia, Germany
- 11:25 - 11:45 Whippet: An example of joint collaboration for accelerated biodiversity research and beyond
Carola Greve, LOEWE-Centre for Translational Biodiversity Genomics, Germany
- 11:45 - 12:00 Q&A
- 12:00 - 12:10 LRUA24 Concluding remarks
Adam Ameur & Olga Vinnere Petterson, SciLifeLab Genomics, Sweden
- 12:10 - 13:00 LUNCH

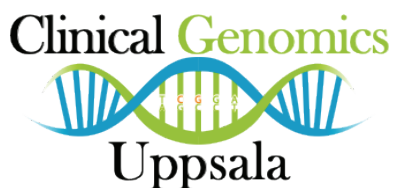
***** CONCURRENT WORKSHOPS *****

See *LRUA24 WORKSHOP PROGRAM* for detailed agendas

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|---------------|--|-----------------------|
| 13:00 - 16:30 | BIODIVERSITY: UNDER THE PLANETARY BIOLOGY UMBRELLA | ROOM: <i>Sal XI</i> |
| 13:00 - 16:00 | CLINICAL GENOMICS | ROOM: <i>Sal X</i> |
| 13:00 - 16:30 | LONG-READ TRANSCRIPTOMICS: WORKFLOW & APPLICATIONS | ROOM: <i>Sal IX</i> |
| 13:00 - 16:20 | REFERENCE GENOME ASSEMBLY: ISSUES AND SOLUTIONS | ROOM: <i>Sal VIII</i> |
| 13:00 - 16:00 | SEQUENCING FACILITY NETWORK:
BEST PRACTICES FOR SEQUENCING OF NON-MODEL ORGANISMS | ROOM: <i>Sal IV</i> |



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