LRUA 24

LONG-READ SEQUENCING UPPSALA

PROGRAM
LRUA24, Oct 21-23 2024

lrua2024.se
Uppsala University Main Buildning, 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 Ameur, Olga Vinnere Petterson & Ida Höijer, 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** *Sponsored by Swedish Society for Medical Genetics*

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 1
Gregor Diensthuber, Venda 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

CHAIR: Susan Kloet

09:00 - 09:30 Decoding the central dogma with single molecules
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, Centro Nacional de Análisis Genómico (CNAG), Spain

10:10 - 10:20 Why the Megaruptor 3 is your best partner for long-read sequencing
Stefan Pellenz, Hologic Diagenode, Germany

10:20 - 10:50 COFFEE & POSTERS

HUMAN/MEDICAL SEQUENCING

CHAIR: Lars Feuk

10:50 - 11:20 High resolution assessment of rare genetic disease
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 *Sponsored by Swedish Society for Medical Genetics*

HUMAN/MEDICAL SEQUENCING continued

CHAIR: Ulf Gyllensten

13:30 - 13:50 Ultra-fast deep-learned CNS tumor classification during surgery
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 Höijer,. 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 demonstation
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 Eberle, PacBio, USA

10:50 - 11:05 Building and training tools for the complete genome
Mike Eberle, 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 *Sponsored by Swedish Society for Medical Genetics*

*** CONCURRENT WORKSHOPS ***

See LRUA24 WORKSHOP PROGRAM for detailed agendas

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

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LRUA24 Organizing Committee: Adam Ameur, Olga Vinnere Pettersson, Ida Höijer, Susanne Hellstedt Kerje, Anna Olsson