LRUA 24

LONG-READ SEQUENCING UPPSALA

WORKSHOP PROGRAM
LRUA24, Oct 21-23 2024

lrua2024.se
Uppsala University Main Buildning, Uppsala, Sweden

WEDNESDAY, OCTOBER 23

LONG-READ TRANSCRIPTOMICS: WORKFLOW & APPLICATIONS

ROOM: Sal IX

Workshop organizers: Ana Conesa & Carolina Monzó, CSIC Valencia

This workshop offers a comprehensive exploration of the long-read RNA sequencing (RNA-seq) workflow, encompassing essential steps from library preparation to advanced applications. LongTREC fellows will present on critical topics including sequencing strategies, data mapping, transcript identification, analysis pipelines, and specialized applications such as genome annotation, single-cell analysis, metatranscriptomics, epitranscriptomics, and ploidy analysis. Attendees will gain valuable insights into the latest methodologies, tools, and best practices in long-read RNA-seq.

13:00 - 13:10 Welcome & Introduction
Ana Conesa, CSIC Valencia, Spain

SESSION 1: Long-Read RNA-Seq Workflow

13:10 – 13:25 Library preparation and sequencing strategies
Satrio Benowo, University of Nottingham, UK
Juan Francisco Cervilla Martinez, Earlham Institute, UK
An in-depth look at Nanopore sequencing, focusing on crucial steps like RNA extraction and library preparation. Satrio and Fran will discuss strategies such as adaptive sampling to optimize sequencing efficiency and address common troubleshooting techniques.

13:25 – 13:40 Mapping
Mahmud Sami Aydın, Stockholm University, Sweden
Sami will address the challenges of mapping long-read RNA sequences, introducing tools like minimap2, deSALT, and uLTRA. The talk will emphasize accurate spliced RNA mapping and efficient computational methods.

13:40 – 13:55 Transcript identification and quantification
Tianyuan Liu, CSIC Valencia & University of Valencia, Spain
Tian will explore tools for identifying and quantifying transcripts using long-read sequencing. The presentation will cover methods for novel transcript discovery and the use of tools like SQANTI3 to enhance transcriptome analysis.

13:55 – 14:20 Analysis pipelines
Fabian Jetzinger, BioBam Bioinformatics, Spain
Yalan Bi, Max Planck Institute for Molecular Genetics, Germany
Ana Conesa, CSIC Valencia, Spain
Fabian, Yalan, and Ana will present comprehensive analysis pipelines tailored for long-read RNA-seq data. They will showcase tools such as IsoTools and OmicsBox for tasks including transcriptome reconstruction, alternative splicing analysis, and pathway enrichment. Ana will present the SQANTI3 pipeline including tappAS, focusing on transcriptome characterization and functional analysis.

14:20 - 14:45 COFFEE BREAK

SESSION 2: Applications of Long-Read RNA-Seq

14:45 – 15:00 Genome annotation
Fabio Zanarello, Centre for Genomic Regulation, Spain
Fabio will discuss enhancing genome annotation using long-read sequencing data. The talk will focus on addressing challenges in annotating both model and non-model organisms and highlight the integration of tools like AUGUSTUS for improved accuracy.

LONG-READ TRANSCRIPTOMICS: WORKFLOW & APPLICATIONS continued

ROOM: Sal IX

15:00 – 15:15 Single-cell transcriptomics
Eamon McAndrew, Institut de Pharmacologie Moléculaire et Cellulaire, France
Eamon will introduce advancements in single-cell long-read transcriptomics. The session will highlight the advantages of full-length transcript coverage in single-cell datasets and discuss scalability challenges and solutions in this emerging field.

15:15 – 15:30 Metatranscriptomics
Carmen Lafuente Sanz, Genoscope-CEA, France
Carmen will examine the application of long-read sequencing in metatranscriptomics. The presentation will focus on accurately mapping transcripts within complex metagenomic datasets and the benefits of capturing rare transcripts with long reads.

15:30 – 15:45 Epitranscriptomics
Leda Katopodi, Centre for Genomic Regulation, Spain
Leda will provide an overview of identifying RNA modifications using long-read sequencing technologies. The talk will review current software tools available for epitranscriptomic analysis and discuss future directions in the field.

15:45 – 16:00 Ploidy analysis
Nadja Nolte, National Institute of Biology, Slovenia
Pablo Angulo Lara, Italian Institute of Technology, Italy
Nadja and Pablo will introduce methods for allele-specific expression and ploidy analysis using long-read RNA-seq. They will discuss tools like LORALS and IDP-ASE for analyzing expression differences at the gene and isoform levels between alleles.

16:00 – 16:25 Open discussion
An opportunity for attendees to engage in an open discussion, ask questions, and share insights.

16:25 – 16:30 Concluding remarks

BIODIVERSITY: UNDER THE PLANETARY BIOLOGY UMBRELLA

ROOM: Sal XI

Workshop organizers: Jacob Höglund, Uppsala University & Anabella Aguilera, Swedish University of Agricultural Sciences

Long reads are required for accurate assembly and annotation of genomes and combined with population genomics and barcoding approaches can provide useful resources for conservation and biodiversity research. In this workshop we will discuss how long-read genomes can be used for assessing both species diversity and genetic diversity.

13:00 - 13:30 Leveraging long-reads for conservation genomics
Jacob Höglund, Uppsala University, Sweden

13:30 - 14:00 Deciphering the complex polyploid genomes and evolution of timothy grasses – *P. nodosum*, *P. alpinum* and *P. pratense*
Pär Ingvarsson, Swedish University of Agricultural Sciences, Sweden

14:00 - 14:30 COFFEE BREAK

14:30 - 15:00 Long-read metabarcoding to describe microbial diversity and patterns of evolution
Mawash Jamy, Swedish University of Agricultural Sciences, Sweden

15:00 - 15:30 Different modes of chromosomal evolution in fungi revealed by long-read sequencing
Helle Tessand Baalsrud, Norwegian University of Life Sciences, Norway

15:30 - 15:35 Short break

15:35 - 16:30 Round table discussion

CLINICAL GENOMICS

ROOM: Sal X

Workshop organizers: Malin Melin & Claes Ladenvall, SciLifeLab Clinical Genomics Uppsala

Long-read sequencing is a powerful tool for analyzing complex genomic features, methylation patterns, and other intricate aspects of the genome. In this workshop we will hear insights from early adopters who are pioneering the use of this technology in healthcare settings and explore what applications are spearheading the introduction of long-read sequencing in the clinic.

13:00 - 13:20	Welcome & Introduction *Malin Melin, SciLifeLab Clinical Genomics, Uppsala University, Sweden*
13:20 - 13:50	A path towards long-read whole genome sequencing in the clinic *Melanie Tanguy, Genomics England, UK*
13:50 - 14:05	Clinical long read whole genome sequencing to confirm diagnosis in neonatal hypotonia *Hans Matsson, Uppsala University Hospital, Sweden*
14:05 - 14:20	Whole genome long-read sequencing in SV calling and variant phasing *Giancarlo Tomio, Center for Cardiovascular Genetics and Gene Diagnostics, Swiss Foundation for People with Rare Diseases, Switzerland*
14:20 - 14:50	**COFFEE BREAK**
14:50 - 15:05	Long-read sequencing as evolving methodology in clinical genetic testing *Hannes Erdmann, Medical Genetics Center Munich & Friedrich-Baur-Institute, Germany*
15:05 - 15:20	Rapid diagnosis of leukemic aberrations using nanopore sequencing *Tatjana Pandzic, Uppsala University Hospital, Sweden*
15:20 - 15:55	Panel discussion
15:55 - 16:00	Concluding remarks

REFERENCE GENOME ASSEMBLY: ISSUES AND SOLUTIONS

ROOM: Sal VIII

Workshop organizers: Kerstin Howe, Wellcome Sanger Institute & Henrik Lantz, SciLifeLab NBIS

All genome projects are different and come with their own challenges. In this workshop we will explore what has been encountered over the recent years in reference genome assembly generation. For the assembly clinic, please bring your own assembly issues to discuss and hopefully resolve!

13:00 - 14:00	Tackling genome assemblies across the Tree of Life: Lessons learned from the first 3,000 species *Shane McCarthy, Wellcome Sanger Institute, UK*
14:00 - 14:20	Phasing or purging: Tackling the genome assembly of a highly heterozygous species in the era of high-accuracy long reads *Nadège Guiglielmoni, Universität zu Köln, Germany*
14:20 - 14:40	Genome assembly of ethanol-preserved specimens with amplified long-read sequencing *Bernhard Bein, Senckenberg Institute for Nature Research, Germany*
14:40 - 15:10	**COFFEE BREAK**
15:10 - 16:20	Assembly clinic *Bring along your examples of assembly issues and/or your questions, to be discussed and hopefully improved with the experience and insight of the people present! Ad hoc presentations are welcomed and encouraged.*

SEQUENCING FACILITY NETWORK: BEST PRACTICES FOR SEQUENCING OF NON-MODEL ORGANISMS

ROOM: Sal IV

Workshop organizers: Olga Vinnere Petterson, SciLifeLab Genomics & Ave Tooming-Klunderud, University of Oslo

How difficult is it to sequence a genome now? Join us to investigate the remaining challenges and discuss how we have tackled (some of) them.

13:00 - 13:30	Long-read sequencing and genome assembly of natural history collection samples and challenging specimens *Carola Greve, Senckenberg Research Institute, Germany*
13:30 - 13:50	Too slimy or too repetitive? Tackling the challenging genome of the cone snail *Conus textile* *Giada Ferrari, University of Oslo, Norway*
13:50 - 14:10	Revving up Revio workflows: Supercharging Sanger's long-read pipelines *Iraad Bronner, Welcome Sanger Institute, UK*
14:10 - 14:30	**COFFEE BREAK**
14:30 - 14:50	Sanger Darwin Tree of Life sharing their experience
14:50 - 15:30	Troubleshooting in the lab while sequencing non-models: Q&A
15:30 - 16:00	Open discussion & closing the session