



Long-Read Sequencing Workshop

Farmington, Connecticut

May 13-16, 2024

Workshop Schedule

All times listed are Eastern Daylight Time (GMT -4)

Monday, May 13th

- 10:30 am Shuttle from Courtyard – Farmington to JAX
- 11:00 am Registration Opens
- 11:30 - 12:45 pm Lunch & Learn: Introduction to Long-Read Sequencing Methods/Outputs
Christine Beck, Ph.D., The Jackson Laboratory
- 12:45 – 1:00 pm Break
- 1:00 pm Opening Remarks

Session 1 – Recent Advances in Long-Reads

- 1:10 pm RNA Isoform regulations across mammalian brain regions, post-natal development, and species – or – single-cell isoforms in time and space
Hagen Tilgner, Ph.D., Weill Cornell Medical Center
- 1:35 pm Studying tumor evolutionary lineages and cell fate trajectories with high throughput single cell RNA sequencing
Ruli Gao, Ph.D., Northwestern University
- 2:00 pm Long-reads at scale for pop-gen and rare disease applications
Kiran Garimella, Ph.D., Broad Institute
- 2:25 pm New pangenome associated computational methods for nanopore sequencing data
Benedict Paten, Ph.D., University of California at Santa Cruz
- 2:50 pm Break

Session 2 – Transcriptomics/Isoforms

- 3:20 pm Visualizing nascent RNA
Karla Neugebauer, Ph.D., Yale University
- 3:45 pm BigBrain: Genetic analysis of novel transcript expression in 11,415 human brain transcriptomes
Kailash Babu Panneerselvam, M.Tech, Icahn School of Medicine at Mount Sinai

- 4:00 pm **Defining aging-associated RNA splicing as an oncogenic factor in breast tissue using long-read RNA sequencing**
Brittany Angarola, Ph.D., The Jackson Laboratory
- 4:15 pm **Isoform-Resolved transcriptome reveals thousands of novel genes in human preimplantation embryos**
Robert Sebra, Ph.D., Icahn School of Medicine at Mount Sinai
- 4:40 pm **Long-read RNA-seq atlas of novel microglia isoforms elucidates disease-associated genetics regulation of splicing**
Jack Humphrey, Ph.D., Icahn School of Medicine at Mount Sinai
- 4:55 pm **Harnessing the template switching activity of UltraMarathonRT to expand the capabilities of RNA-Seq**
Li-Tao Guo, Ph.D., RNAConnect
- 5:10 pm **Dissecting the splicing regulation in single cells: A long-read multiomics journey and data challenges ahead**
Mariela Cortes Lopez, Ph.D., New York Genome Center
- 5:35 pm **Opening Reception**
- 6:00 pm **Welcome Dinner**
- 8:00 pm **Shuttle from JAX to Courtyard – Farmington**

Tuesday, May 14th

- 9:00 am **Shuttle from Courtyard – Farmington to JAX**

Session 3 – Epigenomics

- 9:30 am **New insights into the genetic and epigenetic variation of complete human centromeres**
Glennis Logsdon, Ph.D., University of Pennsylvania
- 9:55 am **Genomic architectures of extrachromosomal DNA circles in tumors revealed by long-read sequencing**
Chia-Lin Wei, Ph.D., University of Washington
- 10:20 am **Multi-omic sequencing: Obtaining the genomes, transcriptome, methylome, and open chromatin in one Revio SMRT Cell**
Aaron Wenger, Ph.D., PacBio, Inc.
- 10:35 am **Break**

11:00 am **KEYNOTE: Transposon genomics from single loci to single cells**
Molly Gale Hammell, Ph.D., New York University

12:00 pm **Lunch**

12:45 pm **Group photo**

Session 3 – Epigenomics (continued)

1:00 pm **Long_Circle_Finder: An algorithm for elucidating long extrachromosomal circular DNA (eccDNA) structures through whole-genome long-read sequencing**
Sarmad Mehmood, Ph.D., University of Alabama at Birmingham

1:15 pm **Multimodal deregulation of imprinted gene HM13 leads to overexpression of the MCTS2 retrogene copy of oncogene MCTS1: a pan-cancer study**
Floris Voorthuijzen, M.S., Ghent University

1:30 pm **Haplotype-resolved view of normal and pathogenic gene regulation**
Andrew Stergachis, M.D., Ph.D., University of Washington

1:45 pm **Resolving the chromatin phenotype of Myotonic Dystrophy Type I with targeted Fiber-seq**
Stephanie Bohaczuk, Ph.D., University of Washington

2:00 pm **Break**

Session 4 – Genome Structure

2:30 pm **Using T2T-scale genome assemblies across the mammalian phylogeny to study chromosome evolution**
Rachel O'Neill, Ph.D., University of Connecticut

2:55 pm **Building tools and resources using recent advances in long-read sequencing**
Peter Audano, Ph.D., The Jackson Laboratory

3:10 pm **Improved genome assemblies resolve complex genomic loci on human Y chromosomes**
Pille Hallast, Ph.D., The Jackson Laboratory

3:25 pm **Analysis of amylase structural variants and exploration of genetic diversity in amylase genes through long-read sequencing**
Feyza Yilmaz, Ph.D., The Jackson Laboratory

3:40 pm **Long-read sequencing in complex genomes**
Evan Eichler, Ph.D., University of Washington

4:05 pm **Vendor Show & Poster Session**

- 6:15 pm Dinner at Butchers & Bakers
- 9:00 pm Shuttle from Butchers & Bakers to Courtyard – Farmington

Wednesday, May 15th

- 9:00 am Shuttle from Courtyard-Farmington to JAX

Session 5 – Long-Read Discoveries in Non-Human Organisms

- 9:30 am **Coordinated Alternative Splicing and Alternative Polyadenylation**
Pedro Miura, Ph.D., University of Connecticut
- 9:55 am **The complete sequence and comparative analysis of ape sex chromosomes**
Kateryna Makova, Ph.D., Pennsylvania State University
- 10:20 am **Segmental duplication – mediated rearrangements alter the landscape of mouse genomes**
Eden Francoeur, B.S., The Jackson Laboratory
- 10:35 am **Break**
- 11:00 am **Efficient characterization of random transgenes with whole genome and adaptive long-read sequencing**
Zachary Freeman, Ph.D., DVM, University of Michigan
- 11:15 am **Returning to full length sequencing of 16S rRNA for community profiling: evaluating sequencing quality of Nanopore reads**
Kendra Maas, Ph.D., University of Connecticut
- 11:30 am **Chromosome-scale Solanum pan-genome reveals the dynamics of paralog evolution**
Michael Schatz, Ph.D., Johns Hopkins University
- 11:55 am **Lunch**
- 1:00 pm **Keynote: Large-scale HiFi-GS in rare disease: Genomic answers for kids**
Tomi Pastinen, M.D., Ph.D., Children's Mercy Kansas City

Session 6 – Clinical Diagnostics

- 2:00 pm **Application of long-read sequencing in clinical oncology**
Arezou Ghazani, M.Sc., Ph.D., FACMG, Brigham and Women's Hospital and Harvard University
- 2:25 pm **Break**

- 2:45 pm **Application of PacBio HiFi long-read genome sequencing in rare developmental disorders**
Liz Werren, Ph.D., The Jackson Laboratory
- 3:00 pm **Characterizing and deciphering structural variations using long-read**
Qihui Li, Ph.D., Johns Hopkins University
- 3:15 pm **Long-read sequencing: From research to clinical**
Fritz Sedlazeck, Ph.D., Baylor College of Medicine
- 3:40 pm **Using long-read sequencing for genomic and epigenomic analysis in patients with Ollier disease and Maffucci syndrome**
Carolina Montano, M.D., Ph.D., FACMG, Johns Hopkins University
- 3:55 pm **Comprehensive genome characterization of childhood T-ALL to discover novel oncogene activation mechanisms**
Petri Pölönen, Ph.D., St. Jude Children's Hospital
- 4:10 pm **Adjourn**
- 4:30 pm **Shuttle from JAX to Courtyard-Farmington**
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Thursday, May 16th – Optional Workshop

Session 7 Workshop - Technical Skills: Long-Read Bioinformatics

**RSVP is requested*

Session Speakers:

Christine Beck, Ph.D., The Jackson Laboratory
Peter Audano, Ph.D., The Jackson Laboratory
Parithi Balachandran, Ph.D., The Jackson Laboratory
Gabriel Rech, Ph.D., The Jackson Laboratory

8:30 am	Shuttle from Courtyard-Farmington to JAX
9:00 am	Introduction to long-reads
9:15 am	PacBio
10:00 am	Break
10:15 am	Oxford Nanopore
11:00 am	Discussion and Questions
11:45 am	Lunch
1:00 pm	Variant Discovery
2:30 pm	Break
2:45 pm	Transcriptomics
3:45 pm	Emerging technologies and other topics (methylation, etc.)
4:30 pm	Discussion, questions, open topics
5:00 pm	Adjourn
5:00 pm	Shuttle from JAX to Courtyard-Farmington

Thursday, May 16th – Optional Workshop

Session 8 Workshop - Technical Skills: Long-Read Technology Library Preparation (Wet Lab Tutorial)

** Separate Registration Required*

8:00 am	Shuttle from Courtyard-Farmington to JAX
8:30 am	Registration
8:50 am	Travel to lab and gather PPE
9:00 am	Introduction/Lab Safety <i>Jennifer Idol, M.S., The Jackson Laboratory</i>
9:15 am	Session 1: High Molecular Weight (HMW) DNA Extraction NEB Monarch HMW DNA Extraction Kit for Cells & Blood <i>Xiaobo Li, Ph.D., The Jackson Laboratory</i>
11:15 am	Lunch
12:15 pm	Travel to lab and gather PPE
12:20 pm	Session 2: PacBio HiFi SMRTBell Library preparation SMRTBell prep Kit 3.0 <i>Yongjun Huang, Ph.D., The Jackson Laboratory</i>
2:20 pm	Break
2:45 pm	Travel to lab and gather PPE
2:50 pm	Session 3: Oxford Nanopore Rapid Barcoding Library prep and sequencing <i>Christine Goldfarb, Ph.D., The Jackson Laboratory</i>
4:50 pm	Q&A/Wrap-Up
5:00 pm	Adjourn
5:00 pm	Shuttle from JAX to Courtyard-Farmington