Leading the search for tomorrow's cures

Workshop on Long-Read Sequencing
September 17 – 20, 2019

Event Schedule

All sessions will be held at the Jackson Laboratory for Genomic Medicine, Farmington, Conn.

Tuesday, September 17th

Session 0: Welcome

8:30 am   Registration Opens

9:00 am   Rachel Goldfeder, Ph.D., Computational Scientist, *The Jackson Laboratory*

9:05 am   Chia-Lin Wei, Ph.D., Director of Genome Technology, *The Jackson Laboratory*

Session 1: Technology Overview

9:15 am   **PacBio Technology Update**
Jonas Korlach, Ph.D., Chief Scientific Officer, *Pacific Biosciences*

9:40 am   James Brayer, Associate Director for Market Development, *Oxford Nanopore*

10:05 am   **Genome Imaging for High Throughput Structural Variation Detection in Cancer and Genetic Disease**
Sven Bocklandt, Ph.D., Director of Scientific Affairs, *Bionano Genomics*

10:30 am   **Highly Scalable Linked Read Library Technology Enables Low Input and Single Tube Library Prep for Haplotype Phasing and De Novo Sequencing**
Tom Chen, Ph.D., Co-Founder & CSO, *Universal Sequencing*

10:40 am   **Long Molecule Phasing With a Single-Tube Short Read Library Prep**
Joseph Mellor, Ph.D., *CEO, seqWell*

10:40 am   Coffee Break

Session 2: Genome Variation

11:30 am   **What's in a Single-Molecule Sequencing Genome?**

Mark Chaisson, Ph.D., Assistant Professor, *University of Southern California*

11:55 am   **Mechanisms of Structural Variation**

Christine Beck, Ph.D., Assistant Professor, *The Jackson Laboratory*

12:20 pm   **100 Genomes in 100 Days: The Structural Variant Landscape in Tomato Genomes**

Michael Schatz, Ph.D., Bloomberg Distinguished Associate Professor, *Johns Hopkins University*

12:45 pm   Lunch

Session 3: Reference Genome

2:00 pm   **Developing Benchmarks for Challenging Variants With Long Reads**

Justin Zook, Ph.D., Human Genomics Team Leader, *National Institute of Standards and Technology*

2:25 pm   **Fish Genome Sequencing With the Help of Tulips**

Christiaan Henkel, Ph.D., Research Scientist, *Norwegian University of Life Sciences*

2:50 pm   **Telomere to Telomere Assemblies of Human Chromosomes**

Karen Miga, Ph.D., Assistant Research Scientist, *UC Santa Cruz Genomics Institute*

3:15 pm   Coffee Break

Session 4A: RNA Biology

4:00 pm   **RNA Biology**

Jacques Banchereau, Ph.D., Director and Professor of Immunological Services, *The Jackson Laboratory*

4:25 pm   **Full-length Alternative Transcript Isoform Analysis Using Nanopore Sequencing**

Angela Brooks, Ph.D., Assistant Professor of Biomolecular Engineering, *University of California, Santa Cruz*

4:50 pm   **Towards High Quality Transcriptome Annotations**

Christopher Vollmers, Ph.D., Assistant Professor, *University of California, Santa Cruz*

Session 4B: Computational Biology

4:00 pm   **Pilot Sequences and Long Read Technologies**

Olgica Milenkov, Ph.D., Professor, *University of Illinois, Urbana-Champaign*

4:25 pm   **Size Matters: Accurate Detection and Phasing of Structural Variations**

Fritz Sedlazeck, Ph.D., Assistant Professor, Human Genome Sequencing Center, *Baylor College of Medicine*

4:50 pm   **Computational Biology**
Yijun Ruan, Ph.D., Professor & Director, JAX Genomic Services, *The Jackson Laboratory*

5:30 pm   Welcome Reception

Wednesday, September 18

Session 5: Cancer

9:00 am   **Telomere Analysis With Long Read Sequencing**
Matthew Meyerson, M.D., Ph.D., Professor of Pathology, *Dana-Farber Cancer Institute*

9:25 am   Chia-Lin Wei, Ph.D., Directo, Genome Technologies, *The Jackson Laboratory*

9:50 am   **3D Genome Organization in Cancer Cells**
Feng Yue, PH.D., Director, Center for Cancer Genomics & Associate Professor of Biochemistry and Molecular Genetics, *Northwestern University*

10:15 am   Coffee Break & Vendor Expo

Session 6: Long Reads in the Clinic

11:30 am   Robert Sebra, Ph.D., Associate Professor, Genetics and Genomics; Director of Technology Development & GCF ,*Icahn School of Medicine at Mount Sinai*

11:55 am   Cecilia Yeung, M.D., Associate Member, *Fred Hutchinson Cancer Research Center*

12:20 pm   **Single Molecule Sequencing for Human Genome Variation: From Repeat Expansions to Whole Chromosome Rearrangements**
Matthew Hestand, Ph.D., Assistant Professor, Division of Human Genetics, *Cincinnati Children's Hospital Medical Center*

12:45 pm   Lunch & Poster Session + Women in Science Networking

Session 7: Genome Assembly

2:25 pm   **Aquila: Diploid Genome Assembly and Variant Detection Based on Linked Reads**
Arend Sidow, Ph.D., Professor, *Stanford University*

2:50 pm   **Telomere-to-Telomere Assembly of a Complete Human X Chromosome**
Adam Phillippy, Ph.D., Investigator, *National Human Genome Research Institute*

Session 8: Selected Abstracts

3:15 pm   **Utility of 10x Genomics Linked-Read Sequencing in Characterizing Short Tandem Repeat Expansion Mutations**
Indhu-Shree Rajan-Babu

3:25 pm   **Nanopore Ultra-Long Read Sequencing Provides Insights Into Structural Variation in Cancer Genome**
Liang Gong

3:35 pm   **Single-Molecule Long-Read Sequencing Reveals Intact RNAs in Sperm**
Yu Sun

3:45 pm   Coffee Break

Session 9A: Microbiome and Infectious Disease

4:15 pm   **Ultra-Deep Long-Read Metagenomics**
Samuel Nicholls, Ph.D., Research Fellow in Bioinformatics, *University of Birmingham*

4:40 pm   **Resolving the Complexity of Human Skin Metagenomes Using Single-Molecule Sequencing**
Julia Oh, Ph.D., Assistant Professor, *The Jackson Laboratory*

5:05 pm   **Microbiome and Infectious Disease**
Ivan Liachko, Ph.D., Founder & CEO, *Phase Genomics*

Session 9B: Epigenetics

4:15 pm   **Using Nanopore Sequencing to Interrogate the Genome, Epigenome and Transcriptome**
Winston Timp, Ph.D., Assistant Professor, *Johns Hopkins University*

4:40 pm   **Illuminating Bacterial Epigenomes to Characterize Pathogens and Microbiome**
Gang Fang, Ph.D., Associate Professor, *Icahn School of Medicine at Mount Sinai*

5:05 pm   **Nanopore Direct RNA Sequencing Enables Comprehensive Transcriptome Profiling and Modification Detection**
Rachel Goldfeder, Ph.D., Computational Scientist, *The Jackson Laboratory*

Session 10: Keynote

5:30 pm   Christopher Mason, Ph.D., Associate Professor, *Weill Cornell Medicine*

6:15 pm   Dinner

Session 11: Technical Skills: Long Read Technology Bioinformatics Analysis (Dry Lab Tutorial)

9:00 am   Introduction & Basic Unix Commands and Cloud Computing

9:15 am   **Analyzing Nanopore sequencing data (base calling, alignment, de novo assembly, SV detection, quality control, data visualization)**
Michael Micorescu**,** Ph.D., *Oxford Nanopore Technologies*

11:00 am   **Nanopolish**
Joanna Pineda, Master of Science Candidate, *Ontario Institute for Cancer Research, University of Toronto*

12:00 pm   **FAIR Data Principles**
Anne Deslattes Mays, Ph.D. Principal Computational Scientist, *The Jackson Laboratory*

12:20 pm   Lunch

1:00 pm   **Bionano Aces: Genome Visualization and Analysis**
Benjamin Clifford, Ph.D., Senior Field Application Scientist, *Bionano Genomics*

2:00 pm   **Analyzing SMRT Sequencing Data** (generating circular consensus reads, alignment, SV detection, IsoSeq (RNA-Seq) mapping and iso-form detection, quality control, data visualization)
Roberto Lleras, Manager, M.S., Manager, Bioinformatics FAS, *Pacific Biosciences*

3:45 pm   Coffee Break

4:15 pm   **SV Detection**
Fritz Sedlazeck, Ph.D., Assistant Professor, *Baylor College of Medicine*

Friday, September 20th

Session 12: Technical Skills: Long Read Technology Library Preparation (Wet Lab Tutorial)

9:00 am   **Introduction**
Jeremy Charette, B.S., Genomic Technologist, *The Jackson Laboratory*

9:30 am   **Extracting High Molecular Weight DNA**
Jennifer Idol, M.S., Genomic Technologist, *The Jackson Laboratory*

12:00 pm   Lunch & presentation (Chris Boles, Ph.D., Sage Sciences)

1:00 pm   **Long Read Library Preparations & PacBio Express Iso-Seq Library Preparation**
Chris Kuhlberg, M.S., Genomic Technologist, *The Jackson Laboratory*

3:00 pm   Coffee Break

3:15 pm   **Sequencing Platforms Overview & Oxford Nanopore Rapid DNA Preparation and Sequencing**
Liang Gong, Ph.D., Postdoctoral Associate, *The Jackson Laboratory*

5:15 pm   Dinner