Workshop on Long-Read Sequencing

2019 Schedule for Planning Purposes Only

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 Oxford Nanopore Technology
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
10:50 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 Using Nanopore Sequencing to Interrogate the Genome, Epigenome and Transcriptome
Winston Timp, Ph.D., Assistant Professor, Johns Hopkins University

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 Deciphering structural complexity and lineage of Double Minutes in Cancer Genomes
Chia-Lin Wei, Ph.D., Director, 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 Emerging Applications for Long Read -Oomics Technologies in Clinical Research
Melissa Smith, Ph.D., Icahn School of Medicine at Mount Sinai

11:55 am Clinical Applications of Nanopore Sequencing in Leukemia
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, Ph.D., MSFHR Postdoctoral Fellow, Department of Medical Genetics, University of British Columbia, and BC Children’s and Women’s Hospital

3:25 pm Nanopore Ultra-Long Read Sequencing Provides Insights Into Structural Variation in Cancer Genome
Liang Gong, Ph.D., Post Doctoral Associate, The Jackson Laboratory for Genomic Medicine

3:35 pm Single-Molecule Long-Read Sequencing Reveals Intact RNAs in Sperm
Yu Sun, PhD candidate, University of Rochester

3:45 pm Group Photo + Coffee Break

Session 9: Keynote

4:15 pm Single-molecule and Planetary-Scale Genomics
Christopher Mason, Ph.D., Associate Professor, Weill Cornell Medicine

Session 10A: Microbiome and Infectious Disease

5:00 pm Ultra-Deep Long-Read Metagenomics
Samuel Nicholls, Ph.D., Research Fellow in Bioinformatics, University of Birmingham

5:25 pm Resolving the Complexity of Human Skin Metagenomes Using Single-Molecule Sequencing
Julia Oh, Ph.D., Assistant Professor, The Jackson Laboratory

5:50 pm Metagenome Assembly Using Proximity Ligation Technology
Ivan Liachko, Ph.D., Founder & CEO, Phase Genomics

Session 10B: Epigenetics
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<tr>
<th>Time</th>
<th>Session Title</th>
<th>Presenter Details</th>
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<tr>
<td>5:00 pm</td>
<td><strong>Genomic and Epigenomic Technologies</strong></td>
<td>Yijun Ruan, Ph.D., Professor &amp; Director, JAX Genomic Services, <em>The Jackson Laboratory</em></td>
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<td>5:25 pm</td>
<td><strong>Illuminating Bacterial Epigenomes to Characterize Pathogens and Microbiome</strong></td>
<td>Gang Fang, Ph.D., Associate Professor, <em>Icahn School of Medicine at Mount Sinai</em></td>
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<td>5:50 pm</td>
<td><strong>Nanopore Direct RNA Sequencing Enables Comprehensive Transcriptome Profiling and Modification Detection</strong></td>
<td>Rachel Goldfeder, Ph.D., Computational Scientist, <em>The Jackson Laboratory</em></td>
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<td>6:15 pm</td>
<td>Dinner</td>
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**Thursday, September 19**

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

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<tr>
<td>9:00 am</td>
<td><strong>Introduction &amp; Basic Unix Commands and Cloud Computing</strong></td>
<td>Rachel Goldfeder, Ph.D., Computational Scientist, <em>The Jackson Laboratory</em></td>
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<td>9:15 am</td>
<td><strong>Analyzing Nanopore sequencing data</strong></td>
<td>Michael Micorescu, Ph.D., <em>Oxford Nanopore Technologies</em></td>
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<td>11:00 am</td>
<td><strong>Nanopolish</strong></td>
<td>Joanna Pineda, Master of Science Candidate, <em>Ontario Institute for Cancer Research, University of Toronto</em></td>
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<td>12:00 pm</td>
<td><strong>FAIR Data Principles</strong></td>
<td>Anne Deslattes Mays, Ph.D. Principal Computational Scientist, <em>The Jackson Laboratory</em></td>
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<td>12:20 pm</td>
<td>Lunch</td>
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<td>1:00 pm</td>
<td><strong>Bionano Access: Genome Visualization and Analysis</strong></td>
<td>Benjamin Clifford, Ph.D., Senior Field Application Scientist, <em>Bionano Genomics</em></td>
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<td>2:00 pm</td>
<td><strong>Analyzing SMRT Sequencing Data</strong></td>
<td>Roberto Lleras, Manager, M.S., Manager, Bioinformatics FAS, <em>Pacific Biosciences</em></td>
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<td>3:45 pm</td>
<td>Coffee Break</td>
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<td>4:15 pm</td>
<td><strong>Structural Variant Detection</strong></td>
<td>Fritz Sedlazeck, Ph.D., Assistant Professor, <em>Baylor College of Medicine</em></td>
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**Friday, September 20th**
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<tr>
<td>9:00 am</td>
<td><strong>Introduction</strong></td>
<td>Jeremy Charette, B.S., Genomic Technologist, The Jackson Laboratory</td>
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<td>9:30 am</td>
<td><strong>Extracting High Molecular Weight DNA</strong></td>
<td>Jennifer Idol, M.S., Genomic Technologist, The Jackson Laboratory</td>
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<td>12:00 pm</td>
<td><strong>Lunch &amp; Presentation</strong></td>
<td>Chris Boles, Ph.D., Sage Sciences</td>
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<td>1:00 pm</td>
<td><strong>Long Read Library Preparations &amp; PacBio Express Iso-Seq Library Preparation</strong></td>
<td>Chris Kuhlberg, M.S., Genomic Technologist, The Jackson Laboratory</td>
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<td>3:00 pm</td>
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<td>3:15 pm</td>
<td><strong>Sequencing Platforms Overview &amp; Oxford Nanopore Rapid DNA Preparation and Sequencing</strong></td>
<td>Liang Gong, Ph.D., Postdoctoral Associate, The Jackson Laboratory</td>
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<td>5:15 pm</td>
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