Long-Read Sequencing Workshop

The Jackson Laboratory for Genomic Medicine
Farmington, Connecticut

April 23-25, 2018

Workshop Schedule

This schedule is subject to change.

Monday, April 23rd, 2018

9:00am **Arrival and Registration**

9:30am **Workshop Overview**
Rachel Goldfeder, Ph.D., Computational Scientist, The Jackson Laboratory for Genomic Medicine

9:45am **Opening Remarks**
Chia-Lin Wei, Ph.D., Director of Genome Technology, The Jackson Laboratory for Genomic Medicine

10:00am **Welcome**
Edison Liu, M.D., President & CEO, The Jackson Laboratory

Session 1: Technology Overview

10:15am **Resolving the Full Spectrum of Human Genome Variation using Linked-Reads**
Stephen Williams, Ph.D., Senior Applications Scientist, 10x Genomics

10:50am **Introduction to PacBio Single Molecule Real Time Sequencing**
Jonas Korlach, Ph.D., Chief Scientific Officer, Pacific Biosciences

11:25am **Detecting Base Detecting Base Modifications Using Nanopore Sequencing**
Winston Timp, Ph.D., Assistant Professor, Johns Hopkins University

12:00pm **Lunch**

Session 2: Applications

1:15pm **3D Genome Mapping Technologies**
Yijun Ruan, Ph.D., Professor and Director of Genome Sciences, The Jackson Laboratory for Genomic Medicine

1:45pm **Extrachromosomal Amplified Oncogenes as Drivers of Glioma Tumor Evolution**
Roel Verhaak, Ph.D., Professor and Associate Director of Computational Biology, The Jackson Laboratory for Genomic Medicine

2:15pm **Group photograph and coffee break**

Session 3: Hands-on Tutorials

2:30 – 5:30pm **Technical Protocols**
10x Genomics Library Preparation

2:30 – 5:30pm **Computing**
Introduction to Amazon Web Services
Oxford Nanopore Technology Data Analysis

6:00pm **Welcome Reception at Butchers and Bakers**
70 Farmington Ave Suite 101, Farmington, CT 06032
By invitation only

Tuesday, April 24th, 2018

Session 4: Resolving Challenging Variants and Complicated Genomic Regions

9:00am **In Pursuit of Perfect Personal Genomes**
Michael Schatz, Ph.D., Associate Professor, Johns Hopkins University

9:30am **Complete Haplotype Assembly of Heterozygous Diploids Using Trios**
Adam Phillippy, Ph.D., Investigator, National Human Genome Research Institute

10:00am **Genome in a Bottle: Integrating Multiple Technologies to Form Benchmark Structural Variants**
Justin Zook, Ph.D., Research Scientist, National Institute of Standards and Technology

10:30am **Resolving Complex Genomic Regions Using 100 kb+ Nanopore Long Reads**
Miten Jain, Ph.D., Postdoctoral Researcher University of California Santa Cruz

11:00am **Break**

Session 5: Genome Technology Development

11:15am **Haplotyping of Key Cardiac Disease Genes Using Long-Read Sequencing-Auditorium**
Alexandra Dainis, Graduate Student, Stanford University

11:45am **Beyond NGS: Bionano Genome Mapping Reveals Structural Variation in Cancer and Genetic Disease**
Sven Bocklandt, Ph.D., Sr. Application Specialist, Bionano Genomics, Inc.

Session 6: Exhibition and Poster Session

12:15pm **Lunch, Expo, and Poster Session** *in the lobby and Helix café*

Session 7: Hands-on Tutorials

1:30 – 5:30pm **Technical Protocols**
Oxford Nanopore Technology Library Preparation
Pacific Biosciences Library Preparation
BioNano Genomics Platform Overview

1:30 – 5:30pm **Computing**
BioNano Genomics Data Analysis
10x Genomics Data Analysis
Pacific Biosciences Data Analysis

5:30pm **Dinner** *in the Helix café*

Wednesday, April 25th, 2018

Session 8: Understanding Disease

9:00am **Long-Read Sequencing Meets Human Genomics**
Kai Wang, Ph.D., Associate Professor, Children's Hospital of Philadelphia

9:25am **3D Genome Structure in Cancer Cells**
Feng Yue, Ph.D., Assitant Professor, Pennsylvania State School of Medicine

9:50am **Understanding Complex Genetics of Human & Infectious Disease Leveraging Recent Advances in NGS and Molecular Technology**
Bobby Sebra, Ph.D., Associate Professor, Director of Technology Development, Iachn Institute, VP of Technology Development at Sema4

10:15am **Diagnosing Genetic Diseases with Next Generation Genome Mapping**
Hayk Berseghyan, Ph.D., Postdoctoral Fellow, Children's National Medical Center

10:40am **Break**

11:00am **Towards a Universal Diagnostic Test for Infection Using Rapid Nanopore Metagenomics**
Justin O'Grady, Ph.D., Associate Professor, Norwich Medical School, University of East Anglia

Session 9: Sample Preparation Strategies

11:25am **Cas9-assisted Sample Preparation Strategies for Long-Read Sequencing**
Chris Boles, Ph.D., Chief Scientific Officer, Sage Science, Inc.

11:50am **Hybridization Capture-Based Target Enrichment for Long-Read Sequencing**
Matt McNeill, Ph.D., Staff Scientist, Integrated DNA Technologies

12:15pm **Lunch**

Session 10: Gene Expression and Epigenetics

1:15pm **Getting the Entire Message: The Full-Length Nature of What Genes are Saying**
Hagen Tilgen, Ph.D., Assistant Professor, Weill Cornell Medicine

1:40pm **Using Long-Read Sequencing to Study RNA Biology**
Brent Graveley, Ph.D., Professor, University of Connecticut

2:05pm **Using SMRT-seq to Interrogate the Epigenetic Landscape in Mammals**
Andrew Xiao, Ph.D., Associate Professor, Yale University of Stem Cell Center

2:30pm **Closing Remarks**
Chia-Lin Wei, Ph.D. and Rachel Goldfeder Ph.D., Genome Technologies, The Jackson Laboratory