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 Korbach, Ph.D., Chief Scientific Officer, Pacific Biosciences

11:25am  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., Assistant 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