

# Biology at True Resolution

2018 ASHG CONFERENCE GUIDE

Visit us at booth #635

# Biology at True Resolution: Enabling a Comprehensive View of Genomics

**WEDNESDAY, OCTOBER 17 | 12:30 PM – 1:45 PM | ROOM 30 C-D, UPPER LEVEL**

Learn how researchers are using groundbreaking technology from 10x Genomics to reveal insights into cellular systems and human genetics:

- Understand genomic diversity
- Reveal genomic heterogeneity
- Profile the chromatin landscape of thousands of individual nuclei
- Perform high resolution genetic screening and enhanced single cell phenotyping of complex cell populations



**MOLLY GASPERINI**

Graduate Student  
University of Washington



**JIMMIE YE, PHD**

Assistant Professor  
University of California, San Francisco

## **COLABS PRESENTATIONS**

### **Getting Started with Single Cell Transcriptomics**

**WEDNESDAY, OCTOBER 17 | 4:00 PM – 4:30 PM | COLABS THEATER | BOOTH #241**

Understand the considerations for experimental design, sample preparation, and downstream data analysis for single cell gene expression experiments.

### **Accelerating Biology: New Product Innovations from 10x Genomics**

**THURSDAY, OCTOBER 18 | 10:30 AM – 11:00 AM | COLABS THEATER | BOOTH #241**

Meet our latest innovations, each transforming our understanding of biology from DNA, RNA to epigenetics, cell by cell.

### **Unraveling Multiple Dimensions of Cellular Heterogeneity**

**THURSDAY, OCTOBER 18 | 1:45 PM – 2:15 PM | COLABS THEATER | BOOTH #241**

Learn how to examine cellular heterogeneity and analyze the complexity of biological systems, both within a population and over time, all at the single cell level.

### **Visualization of Multiple Single Cell Genomic Datatypes**

**FRIDAY, OCTOBER 19 | 1:45 PM – 2:15 PM | COLABS THEATER | BOOTH #241**

Deepen your understanding of complex cell populations with our easy to use software and visualization tools.

## **SCIENTIFIC SESSIONS**

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### **Understanding Tumor Heterogeneity from Single Cell Sequencing of Genomes, Transcriptomes and Epigenomes**

**WEDNESDAY, OCTOBER 17 | 10:30 AM – 12:30 PM | BALLROOM 20A - UPPER LEVEL**

**CONCURRENT INVITED SESSION I**

Xinying Zheng **10X** GENOMICS

Elham Azizi, Memorial Sloan Kettering Cancer Center

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### **What are We Missing? Identification of Previously Underappreciated Mendelian Variants**

**THURSDAY, OCTOBER 18 | 11:00 AM – 12:30 PM | BALLROOM 20A - UPPER LEVEL**

**CONCURRENT PLATFORM SESSION E**

Daniel Koboldt, Nationwide Children's Hospital

Zeynep Coban Akdemir, Baylor College of Medicine

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### **Single Cell Approaches to Reveal Disease Biology**

**THURSDAY, OCTOBER 18 | 11:00 AM – 12:30 PM | BALLROOM 20BC - UPPER LEVEL**

**CONCURRENT PLATFORM SESSION E**

Kimberly Aldinger, Seattle Children's Research Institute

Manolis Kellis, Massachusetts Institute of Technology

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### **Increasing Functional Resolution Through Single Cell Analysis**

**SATURDAY, OCTOBER 20 | 9:45 AM – 11:15 AM | ROOM 6B - UPPER LEVEL**

**CONCURRENT PLATFORM SESSION H**

Nick Banovich, Translational Genomics Research Institute

Mingyao Li, University of Pennsylvania

## POSTERS

### Wednesday, October 17th

#### 2:00 PM – 3:00 PM

A novel next generation sequencing based genetic test platform for congenital adrenal hyperplasia due to CYP21A2 defects

*Qizong Lao | #2877  
NIH*

#### 2:00 PM – 3:00 PM

Haplotype resolved analysis and improved genome coverage with Linked-Reads

*Stephen Williams | #385*



#### 2:00 PM – 3:00 PM

Classification of prostate cancer patients as indolent or aggressive using machine learning approach

*Tarun karthik kumar Mamidi | #825  
Louisiana State University*

#### 2:00 PM – 3:00 PM

Characterizing tumor heterogeneity at single cell level

*Claudia Catalanotti | #903*



#### 3:00 PM – 4:00 PM

SAMPLLOT: Rapid structural variant visualization for short, long, linked, and phased reads

*Ryan Layer | #1446  
University of Colorado, Boulder*

#### 3:00 PM – 4:00 PM

A public benchmarking resource of 194,731 CNV calls generated from a combination of short reads, linked long reads and true long read technologies

*Xiao Chen | #1638  
Illumina*

#### 3:00 PM – 4:00 PM

Single nuclei RNA-seq to decipher hypothalamic transcriptional response to chronic topiramate administration

*Kathy Ushakov | #2202  
Boston Children's Hospital*

#### 3:00 PM – 4:00 PM

Using linked-read sequencing to resolve pathogenic mutations and overcome pseudo-gene interference in PKD1 for autosomal dominant polycystic kidney disease testing

*Charlly Kao | #2874  
Children's Hospital of Philadelphia*

#### 3:00 PM – 4:00 PM

Classification of cell type from single-cell RNA sequencing data by deep neural networks

*Charolyn Lou | #3534  
University of Pennsylvania*

#### 3:00 PM – 4:00 PM

Detecting inversion polymorphisms at population scale with linked read sequencing

*Christopher Whelan | #394  
Broad Institute*

#### 3:00 PM – 4:00 PM

New methods for discovery and interpretation of allelic diversity in human genomes

*Robert Fulton | #442  
Washington University in St. Louis*

#### 3:00 PM – 4:00 PM

Gene regulation and cell-cell communication underlying intra-tumoral heterogeneity in HCC liver cancer

*Bojan Losic | #852  
Icahn Institute for Genomics and Multiscale Biology*

#### 3:00 PM – 4:00 PM

Genetic heterogeneity profiling by single cell RNA sequencing

*Zilu Zhou | #894  
University of Pennsylvania*

#### 3:00 PM – 4:00 PM

A comprehensive cross-platform comparison of single-cell RNA sequencing using reference samples

*Charles Wang | #924  
Loma Linda University*

## POSTERS

### Thursday, October 18th

#### 2:00 PM – 3:00 PM

Imputation of single-cell gene expression with an autoencoder neural network

Rui Li | #1687

University of Idaho

#### 2:00 PM – 3:00 PM

Single cell sequencing of iPSC neural cells from Down syndrome patients uncovers perturbed cell differentiation

Joakim Klar | #1747

Science for Life Laboratory

#### 2:00 PM – 3:00 PM

Supporting single-cell RNA-seq at scale in the genomics platform at the Broad Institute

Cole Walsh | #1783

Broad Institute

#### 2:00 PM – 3:00 PM

Single cell analysis of Alzheimer's disease

Manolis Kellis | #1897

Massachusetts Institute of Technology

#### 2:00 PM – 3:00 PM

Combining single-cell RNA and DNA sequencing analysis to measure genomic instability at individual clone resolution in gastric cancer

Sue Grimes | #817

Stanford University

#### 2:00 PM – 3:00 PM

Linked read technology enables disambiguation of structural variants to PMS2 and PMS2CL

Jeroen Van den Akker | #919

Color Genomics, Inc.

#### 3:00 PM – 4:00 PM

LinkedSV: Detection of mosaic structural variants from linked-read sequencing

Li Fang | #1486

Children's Hospital of Philadelphia

#### 3:00 PM – 4:00 PM

Detection of structural variants using linked-reads with novel algorithms

Ian Fiddes | #1564

10X GENOMICS

#### 3:00 PM – 4:00 PM

How well can you detect structural variants: towards a standard framework to benchmark human structural variation

Justin Zook | #1648

National Institute of Standards and Technology

#### 3:00 PM – 4:00 PM

Cell-type specific differential gene expression in post-mortem snRNA-seq of the depressed brain

Malosree Maitra | #1744

Douglas Institute

#### 3:00 PM – 4:00 PM

High-throughput single nuclei RNA-seq from limited amounts of frozen human and mouse tissues

Anugraha Raman | #1780

Ludwig Institute for Cancer Research

#### 3:00 PM – 4:00 PM

Studies of liver tissue identifies functional gene regulatory elements associated to gene expression, type 2 diabetes and other metabolic diseases

Claes Wadelius | #2188

Science for Life Laboratory

#### 3:00 PM – 4:00 PM

De novo human genome assemblies reveal novel sequences in diverse populations

Karen Wong | #440

University of California, San Francisco

## POSTERS

### Friday, October 19th

#### 2:00 PM – 3:00 PM

Single-nucleus RNA-sequencing of human subcutaneous adipose tissue reveals a heterogeneous population of 12 cell types

Marcus Alvarez | #1709

University of California, Los Angeles

#### 2:00 PM – 3:00 PM

Single-cell RNA-sequencing of bulk peripheral blood mononuclear cells from idiopathic multicentric Castleman disease (iMCD) reveals leukocyte heterogeneity underlying disease pathology

Michael Gonzalez | #2279

The Children's Hospital of Philadelphia

#### 2:00 PM – 3:00 PM

Building human reference genomes for Africa

Martin Pollard | #2771

Wellcome Sanger Institute

#### 2:00 PM – 3:00 PM

Beyond the exome: Identification of novel mutational signatures and oncogenic dependencies in the non-coding genome of multiple myeloma

Cody Ashby | #899

University of Arkansas for Medical Sciences

#### 3:00 PM – 4:00 PM

Single cell transcriptomes of sickle cell disease PBMC identifies markers of disease severity in distinct cell populations

Samuel Lessard | #1004

Bioverativ

#### 3:00 PM – 4:00 PM

Precise breakpoint detection of balanced and unbalanced structural variation in whole genome sequencing data using haplotype blocks created by linked-reads

Jeroen Knijnenburg | #2990

Leiden University Medical Center

## LEARN MORE

### Getting Started Guide

Whether you have questions about how to design your experiments, optimize parameters, or identify the computational/analytical tools necessary to best analyze your single cell gene expression data, this guide has you covered. Take full advantage of the rich information enabled by single cell transcriptomic technology by downloading the getting started guide today!

[Download the Guide](#)

