

# KEYSTONE SYMPOSIA

on Molecular and Cellular Biology

## Beyond a Million Genomes: From Discovery to Precision Health (A4)

January 21-25, 2020 • Beaver Run Resort • Breckenridge, Colorado, USA

Scientific Organizers: Alan R. Shuldiner, Dan Roden and Elaine R. Mardis

Supported by the Directors' Fund

Abstract & Scholarship Deadline: September 26, 2019 / Abstract Deadline: October 22, 2019 / Discounted Registration Deadline: November 21, 2019

### TUESDAY, JANUARY 21

#### Arrival and Registration

### WEDNESDAY, JANUARY 22

#### Welcome and Keynote Address

**Eric S. Lander**<sup>†</sup>, Massachusetts Institute of Technology, USA  
*Talk Title to be Announced*

#### Undiagnosed and Underdiagnosed Mendelian Diseases

**Wendy K. Chung**, Columbia University, USA  
*Monogenic Diseases in the Era of Whole Exome Sequencing*

**Kevin A. Strauss**, Clinic for Special Children, USA  
*Plain People and Precision Medicine*

**Lisa Bastarache**, Vanderbilt University Medical Center, USA  
*EHR Phenomics and Underdiagnosed Mendelian Diseases*

#### Short Talk(s) Chosen from Abstracts

#### Genetic Variation to Mechanisms of Human Disease

**Daniel G. MacArthur**, Broad Institute, USA  
*The Genome Aggregation Database (gnomAD)*

**Eimear E. Kenny**, Icahn School of Medicine at Mount Sinai, USA  
*Using Cryptic Relatedness for Gene Discovery*

**Douglas M. Fowler**, University of Washington, USA  
*Multiplex Experimental Approaches to Understanding Genetic Variation*

#### Short Talk(s) Chosen from Abstracts

#### Poster Session 1

### THURSDAY, JANUARY 23

#### Genome and EHR-Derived Phenomes in Large Patient Populations: Discovery and Implementation of Precision Health

**Catherine Sudlow**, University of Edinburgh, UK  
*UK Biobank*

**Stephanie Devaney**, National Institutes of Health, USA  
*All of Us*

**Zhengming Chen**, University of Oxford, UK  
*China Kadoorie Biobank*

**David H. Ledbetter**, Geisinger Health, USA  
*Geisinger's Genomics and Precision Health Imperative*

#### Short Talk(s) Chosen from Abstracts

#### Panel: Equity in Health and Healthcare: Considerations of Race/Ethnicity in Precision Medicine

**Esteban Gonzalez Burchard**, University of California, San Francisco, USA

**David H. Ledbetter**, Geisinger Health, USA

#### Challenges of Big Data Integration in Genomics and Precision Medicine

**Gonçalo Abecasis**, University of Michigan, USA  
*TOPMED*

**David Glazer**, Verily Life Sciences, USA  
*Talk Title to be Announced*

**Garry P. Nolan**, Stanford University, USA  
*Multidimensional Data Approaches to Characterize the Tumor Microenvironment*

#### Short Talk(s) Chosen from Abstracts

#### Poster Session 2

### FRIDAY, JANUARY 24

#### Translating Cancer Genomics to Clinical Practice

**Elaine R. Mardis**, Nationwide Children's Hospital, USA  
*Integrating Genomics into Precision Cancer Medicine*

**David Hyman**, Memorial Sloan Kettering Cancer Center, USA  
*Talk Title to be announced*

**Gerald P. Linette**, University of Pennsylvania, USA  
*Personalized Cancer Vaccines*

**Katherine Janeway**, Dana-Farber Cancer Institute, USA  
*Talk Title to be Announced*

#### Short Talk(s) Chosen from Abstracts

#### Round Table Discussion: Academia – Industry Collaboration to Enable Genomic Discovery and Implementation of Precision Medicine

**Alan R. Shuldiner**, Regeneron Pharmaceuticals, Inc, USA

**Martin G. Reese**, Fabric Genomics, USA

**Andrew S. Plump**, Takeda Pharmaceuticals, USA

#### Poster Session 3

### SATURDAY, JANUARY 25

#### Pharmacogenomics: Beyond Single Large Effect Size Variants and Implementation

**Dan M. Roden**, Vanderbilt University School of Medicine, USA  
*Challenges and Opportunities in Implementing Pharmacogenomics*

**Marie-Pierre Dubé**, Pharmacogenomics Centre at the Montreal Heart Institute, Canada  
*Gaining Precision with Cardiovascular Clinical Trials Using Genomics*

**Esteban Gonzalez Burchard**, University of California, San Francisco, USA

*Making Precision Medicine Socially Precise*

**Wasun Chantratita**, Mahidol University, Thailand  
*National Program to Eradicate Stevens Johnson Syndrome*

#### Short Talk(s) Chosen from Abstracts

#### Beyond Human Sequence Variation

**Beth Drolet**, Medical College of Wisconsin, USA  
*Mosaicism*

**Nada Jabado**, McGill University Health Centre, Canada  
*Epigenetics of Pediatric Brain Cancer*

**Michael P. Snyder**, Stanford University School of Medicine, USA  
*The Exposome*

#### Short Talk Chosen from Abstracts

#### Meeting Wrap-Up: Outcomes and Future Directions (Organizers)

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**SUNDAY, JANUARY 26**

**Departure**