

# 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, CO, USA

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

Sponsored by Editas Medicine and Novo Nordisk A/S

Discounted Abstract & Scholarship Deadline: October 10, 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

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

**Sir Peter Donnelly**, University of Oxford and Genomics plc, UK  
*Using Genomics to Understand an Individual's Risk for Common Diseases*

#### Undiagnosed and Underdiagnosed Mendelian Diseases

\*Eimear E. Kenny, Icahn School of Medicine at Mount Sinai, USA

**Wendy K. Chung**, Columbia University, USA  
*Horses Morphing into Zebras: Hundreds of Rare Monogenic Diseases Masquerading As Common Diseases*

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

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

**Bryce A. Schuler**, Vanderbilt University Medical Center, USA  
*Short Talk: Identifying Undiagnosed Alpha-1 Antitrypsin Deficiency and Environmental Disease Modifiers via Phenotype Risk Score*

**Margot A. Cousin**, Mayo Clinic, USA  
*Short Talk: Patient-Specific Antisense Oligonucleotide Therapeutic Development for a Girl with Mucopolidosis*

#### Genetic Variation to Mechanisms of Human Disease

\*Wendy K. Chung, Columbia University, USA

**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*

**Monkol Lek**, Yale School of Medicine, USA  
*Genetic Diagnosis to Custom Genetic Therapy*

**Andrew M. Glazer**, Vanderbilt University Medical Center, USA  
*Short Talk: High-Throughput Functional Studies of SCN5A Variants*

**Amrita Basu**, University of California, San Francisco, USA  
*Short Talk: Discovering Biomarkers of Extreme Resistance in High-Risk Breast Cancer*

#### Poster Session 1

### THURSDAY, JANUARY 23

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

\*Dan M. Roden, Vanderbilt University Medical Center, USA

**Kathleen Barnes**, University of Colorado Denver, USA  
*Implementing Personalized Medicine in the Academic Medical Setting: The Colorado Center for Personalized Medicine Experience*

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

**Lili Milani**, University of Tartu, Estonia

*Personalized Medicine Based on Common and Rare Genetic Variants in Estonia*

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

**Manuel A. Rivas**, Stanford University, USA  
*Short Talk: Global Meta-Analysis of UK Biobank, Million Veterans Program, and Biobank Japan*

**Todd Edwards**, Vanderbilt University Medical Center, USA  
*Short Talk: Leveraging T2D GWAS with 1.3M Subjects and Large EHR Resources to Identify and Evaluate Drugs for Repurposing*

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

\*Nishadi Rajapakse, National Institutes of Health, USA

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

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

**Melinda C. Aldrich**, Vanderbilt University Medical Center, USA

**Digna R. Velez Edwards**, Vanderbilt University Medical Center, USA

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

\*Philip F. Smith, NIDDK, National Institutes of Health, USA

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

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

**Garry P. Nolan**, Stanford University, USA  
*Pathology from the Molecular Scale on Up*

**Josh Bonkowsky**, University of Utah, USA  
*Short Talk: Pediatric Personalized Population Health: Integrating Genomic Testing and Data with Longitudinal Pediatric Clinical Outcomes and Treatments in a Free-Standing Children's Hospital*

**Alex Forrest-Hay**, Metabolon Inc., USA  
*Short Talk: Use of Untargeted Metabolomics to Categorize Disease Risk in Human Health Studies Accelerates Precision Medicine and Drug Development*

#### Poster Session 2

### FRIDAY, JANUARY 24

#### Translating Cancer Genomics to Clinical Practice

\*Martin G. Reese, Fabric Genomics, USA

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

**Dejan Juric**, Massachusetts General Hospital, USA  
*Talk Title to be Announced*

**Katherine Janeway**, Dana-Farber Cancer Institute, USA  
*Defining the Clinical Impact of Bringing Genomics to the Pediatric Oncology Clinic*

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

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**Xiaoyan Jia**, University of Michigan, USA

*Short Talk: Comprehensive Functional Classification of Lynch Syndrome Missense Variants*

**Emily Groopman**, Columbia University, USA

*Short Talk: Clinical Utility of Secondary Findings for Hereditary Cancer Predisposition*

### 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

**Pakito Martinez**, Metabolon, USA

**Patrick Kleyn**, Rhythm Pharmaceuticals, USA

### Keynote Address

\***Elaine R. Mardis**, Nationwide Children's Hospital, USA

**Michael P. Snyder**, Stanford University School of Medicine, USA  
*Big Data and Health*

### Poster Session 3

#### SATURDAY, JANUARY 25

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

\***Aaron Pawlyk**, Eunice Kennedy Shriver National Institute of Child Health and Human Development, USA

**Dan M. Roden**, Vanderbilt University Medical Center, USA

*Challenges and Opportunities in Implementing Pharmacogenomics*

**Wasun Chantratita**, Mahidol University, Thailand

*National Program to Eradicate Stevens Johnson Syndrome from Thailand to Asian Countries*

**Esteban E. Burchard**, University of California, San Francisco, USA  
*Making Precision Medicine Socially Precise*

**Katelyn Neely**, Vanderbilt University, USA

*Short Talk: CYP2C19 Phenotype and Adverse Events During Sertraline Treatment in Children and Adolescents*

### Workshop

\***Emily Groopman**, Columbia University, USA

\***Katelyn Neely**, Vanderbilt University, USA

**Andrew R. Marderstein**, Weill Cornell Medicine, USA

*Leveraging Phenotypic Variability to Identify Genetic Interactions and Improve Prediction of Complex Traits*

**Angela Mo**, Georgia Institute of Technology, USA

*African Ancestry Proportion Influences Ileal Gene Expression in Inflammatory Bowel Disease*

**Arbel Harpak**, Columbia University, USA

*Variable Prediction Accuracy of Polygenic Scores within an Ancestry Group*

**Jacob M. Keaton**, Vanderbilt University Medical Center, USA

*Phenome-Wide Association Study of Biogeographic Ancestries Builds an Atlas of Racial Health Disparities*

**Ayesha Muhammad**, Vanderbilt University, USA

*A High-Throughput Cell-Surface Trafficking Assay of KCNE1 Can Identify Functionally Deleterious Variants*

**Megan Lynch**, University of Maryland, Baltimore, USA

*Assessing the Burden of Pathogenic Variants in ACMG Clinically Actionable Genes in the Amish*

### Beyond Human Sequence Variation

\***Christa L. Martin**, Geisinger Health, USA

**Beth Drolet**, University of Wisconsin, USA  
*Mosaicism*

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

*Beyond Identification of Human Sequence Variation: Genome-Informed Therapeutic Development and the Implementation of Population Precision Health*

**Digna R. Velez Edwards**, Vanderbilt University Medical Center, USA

*Short Talk: Association of Genetically-Predicted Placental Gene Expression with Adult Obesity and Blood Pressure Traits*

**Wen Gu**, University of Texas Southwestern Medical Center, USA

*Short Talk: Investigating the Mechanism and Consequences of 2-Hydroxyglutarate Accumulation in Patients with Neurological Abnormalities*

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

#### SUNDAY, JANUARY 26

### Departure