TUESDAY, JANUARY 21
Arrival and Registration

WEDNESDAY, JANUARY 22
Welcome and Keynote Address
*Alan R. Shuldiner, Regeneron Pharmaceuticals, Inc, USA
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: Discovering Biomarkers of Extreme Resistance in Amrita Basu, University of California, San Francisco, USA

Short Talk: High-Throughput Functional Studies of SCN5A Variants Andrew M. Glazer, University of Washington, USA
Genetic Diagnosis to Custom Genetic Therapy
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

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
Poster Session 2

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, Mayo Clinic, USA
Outcomes and Treatments in a Free-Standing Children’s Hospital

Josh Bonkowsky, University of Utah, 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

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

For the most up-to-date details, visit https://www.keystonesymposia.org
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

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

*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
Organ With A View; Birthmarks as a Model of 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