



One Million Genomes: From Discovery to Health

June 4–8, 2018 | Herrenhausen Palace | Hannover | Germany

Scientific Organizers:

Geoffrey S. Ginsburg, Duke University, USA

Teri Manolio, National Institutes of Health, USA

Patrick Boon Ooi Tan, Genome Institute of Singapore, Singapore

In cooperation with Volkswagen Foundation

The completion of the Human Genome Project in 2003 has catalyzed innovations in scientific research and in health care embodied in the term “precision medicine.” Across the globe, many nations are investing in large-scale national sequencing cohort programs resulting in over one million human genomes sequenced and linked to dense phenotypic and clinical data. This Keystone Symposia conference will bring together scientists and leaders from healthcare and industry to discuss how to maximize the value of that investment for human health. It will assemble a unique and highly interdisciplinary international community to articulate how best to use these data-rich resources to provide novel insights into the biology of disease, tools for the management of patients and population health management strategies. The meeting will highlight challenges and potential solutions for germ-line and somatic sequencing programs and make recommendations for optimizing their impact on global health. The crucial role of free and open sharing and exchange of human variation data from these programs in allowing all of them to interpret novel variants and use them in clinical care will be emphasized. The meeting will focus on various scientific challenges for the field, including implementation science, the scalable data infrastructures and analyses required for impact on discovery and clinical care, and the value proposition for the investments that have been made in national programs. The leading edge of clinical impact of clinical sequencing with will be highlighted in sessions on pharmacogenomics, and in developing novel therapeutics.

Session Topics:

- Large-Scale National Sequencing Programs: Implementation to Impact
- Somatic Sequencing Programs: Biological Insights and Diagnosis
- Implementation Science for Genomic and Precision Medicine
- International Data Resources Enabling Genomic Medicine
- The Actionable Genome
- The Value Proposition for National Sequencing
- Pharmacogenomics: The Leading Edge of Genomics Impact in Medicine
- Use of Large-Scale Bio-Clinical Resources for Drug Discovery

Global Health Travel Award Deadline: January 9, 2018

Scholarship Application & Discounted Abstract Deadline: February 7, 2018

Abstract Deadline: March 7, 2018

Discounted Registration Deadline: April 11, 2018



Note: Scholarships are available for graduate students and postdoctoral fellows and are awarded based on the abstract submitted. Submitting an abstract is an excellent opportunity to gain exposure for your work. Global Health Travel Awards are for LMIC investigators. Abstracts submitted by the abstract deadline will also be considered for short talks on the program.

Upper image of GeneChip loaded with hybridized RNA courtesy of National Institute of Arthritis, Musculoskeletal and Skin Diseases, NIH and photographer Rhoda Baer

Meeting Hashtag: #KS1Mgenome
www.keystonesymposia.org/18G1

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KEYSTONE SYMPOSIA

on Molecular and Cellular Biology

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MONDAY, JUNE 4

Arrival and Registration

TUESDAY, JUNE 5

Welcome and Keynote Address

***Geoffrey S. Ginsburg**, Duke University, USA

Sandi Deans, National Health Service England, UK
Delivering Genomic Medicine at a Population Level Across a Health System – The NHS Experience

Large-Scale National Sequencing Programs: Implementation to Impact

***Geoffrey S. Ginsburg**, Duke University, USA

Zhengming Chen, University of Oxford, UK
China Kadoorie Biobank of 0.5 Million People: Opportunities

Andres Metspalu, University of Tartu, Estonia
From Biobanking to Precision Medicine

Gad Rennert, Rappaport Faculty of Medicine, Technion – Israel
Institute of Technology, Israel
Clalit's National Israeli 100K Genomes Personalized Medicine RCT

J. Michael Gaziano, VA Boston Healthcare System, USA
The Million Veteran Program: A Modern Mega-Cohort within a Large Healthcare System – Progress, Challenges and Future Directions

Mark Caulfield, William Harvey Research Institute, Queen Mary
University of London, UK
Genomics England: The 100,000 Genomes Project

Poster Session 1

Somatic Sequencing Programs: Biological Insights and Diagnosis

Peter Campbell, Wellcome Trust Sanger Institute, UK
Precision Oncology through International Collaboration and Data Sharing

Pawel Stankiewicz, Baylor College of Medicine, USA
New Mutation, Mosaicism and Human Disease Traits

***Patrick Boon Ooi Tan**, Biomedical Research Council, Agency for
Science Technology and Research, Singapore
*Environment, Lifestyle and Genetics: Asian Perspectives and
Implications for Precision Medicine*

Adria Jaume Roura Canalda, Nencki Institute of Experimental
Biology of Polish Academy of Sciences, Poland
*Short Talk: Somatic Mutation History of Glioblastoma Patients with
Recurrent Tumors*

WEDNESDAY, JUNE 6

Implementation Science for Genomic and Precision Medicine

***Teri A. Manolio**, National Human Genome Research Institute, USA

Robyn L. Ward, University of Queensland, Australia
Genomic Test Evaluation Frameworks: A Review

John EL Wong, National University Health System, Singapore
*Challenges of Precision Medicine Implementation in a High Volume
Clinical Environment*

Dana C. Crawford, Case Western Reserve University, USA
*Short Talk: Participating in and Return of Results from Precision
Medicine Research: A Survey of Diverse Participants from a Public
Hospital in the United States*

Kathryn North, University of Melbourne, Australia
*Implementing Genomics into Healthcare: A National and Global
Perspective*

Geoffrey S. Ginsburg, Duke University, USA
*A National Network for Implementing Genomic Medicine in Practice
(IGNITE)*

Workshop: Rapid Interpretation of Genomes and Variant Calling

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

Atsuko Imai-Okazaki, National Center for Global Health and
Medicine, Japan
*Incorporation of Originally-Developed Statistical Genetics Methods into
a Clinical Sequencing Platform to Improve Efficiency and Quality of
Variant Annotation*

Peter Bauer, Centogene AG, Germany
Clinical Benefit of Whole Genome Sequencing

Lina Ghaloul Gonzalez, University of Pittsburgh School of Medicine,
USA
*Novel Genomics Applications Reveal the Healthcare Eclipse and
Lead to Better Clinical Outcomes*

Natalie Jäger, German Cancer Research Center, Germany
*Next-Generation Personalized Medicine for High-Risk Paediatric
Cancer Patients - The INFORM Registry*

***Francisco M. De La Vega**, Fabric Genomics, Inc., USA
*You Can't Interpret What You Don't See: Variant Calling from NGS
Data for Clinical Applications*

International Data Resources Enabling Genomic Medicine

***Kathryn North**, University of Melbourne, Australia

Daniel G. MacArthur, Broad Institute, USA
Databases for Rare Disease

Barbara C. Biedermann, University of Basel, Switzerland
*Short Talk: COBEDIAS Empiric Clinical Profiling - A Digital Tool to
Phenotype Disease*

Kristjan Metsalu, University of Tartu, Estonia
Estonian Healthcare Information System

THURSDAY, JUNE 7

The Actionable Genome

***Geoffrey S. Ginsburg**, Duke University, USA

Heidi Rehm, Harvard Medical School, USA
The Clinical Genome Resource

James S. Ware, Imperial College London, UK
Variation in Cardiac Disease Genes: Location Matters

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John R. Giudicessi, Mayo Clinic, USA

Short Talk: Synergistic Use of Rare Disease and Public Exome Databases Question the Nature of Minor Non-Syndromic Long QT Syndrome-Susceptibility Gene-Disease Relationships

Teri A. Manolio, National Human Genome Research Institute, USA
NHGRI/NIH Programs Developing Clinically Actionable Genomic Information

Jill M. Hagenkord, Color Genomics, Inc., USA

Universal Screening for the CDC Tier 1 Genomic Conditions: Opportunities for Health Systems, Government Health Agencies and Sequencing Initiatives

Franziska Singer, ETH Zurich, Switzerland

Short Talk: The Swiss Molecular Tumor Board: Comprehensive Molecular Cancer Diagnostics in the Clinics

Poster Session 2

The Value Proposition for National Sequencing

***Maja Mockenhaupt**, Universitätsklinikum Freiburg, Germany

Lotte Steuten, Fred Hutchinson Cancer Research Center, USA
Multiplex NGS Testing in Cancer Care: An Economic Perspective on Hope, Hype and Value

Sarah Wordworth, University of Oxford, UK

The Health Economic Evidence for Whole Genome Sequencing

Surakameth Mahasirimongkol, Ministry of Public Health, Thailand
Genomics Thailand: Precision Clinical Care on a National Scale

Nusara Satproedprai, Medical Life Science Institute, Thailand
Short Talk: Genomics Thailand: Human Genome Database for Improvement of Health and Medical Services of Thailand

Brett Doble, University of Oxford, UK

Short Talk: Using Routinely Collected "Big Data" to Estimate Healthcare Costs for Rare Disease Patients: An Early Analysis of the UK 100,000 Genomes Project

FRIDAY, JUNE 8

Pharmacogenomics: The Leading Edge of Genomics Impact in Medicine

***Robyn L. Ward**, University of Queensland, Australia

Henk-Jan Guchelaar, Leiden University, Netherlands
The Ubiquitous Pharmacogenomics Project

Mary V. Relling, St. Jude Children's Hospital, USA
Clinical Implementation of Pharmacogenetics

Gabor Marth, University of Utah, USA

Short Talk: Precision Oncology of Advanced Disease: Insights into Refractory and Metastatic Cancers

Maja Mockenhaupt, Universitätsklinikum Freiburg, Germany
Using Genetics to Prevent Stevens Johnson Syndrome/Toxic Epidermal Necrolysis

Jianjun Liu, Genome Institute of Singapore, Singapore
The SaPHIRE Program – Enabling Adverse Drug Reaction Research in Real Time

Folefac Aminkeng, National University of Singapore/ Agency for Science, Technology & Research, Singapore

Short Talk: A missense variant of HFE is associated with Bleomycin-induced Lung Injury in South East Asian Hodgkin Lymphoma Patients

Use of Large-Scale Bio-Clinical Resources for Drug Discovery

***Henk-Jan Guchelaar**, Leiden University, Netherlands

Alan R. Shuldiner, Regeneron Pharmaceuticals, Inc, USA
Discovery of Novel Drug Targets: High-Throughput Genomics of 250,000 Exomes

Carolina Haefliger, AstraZeneca, Sweden

Transforming Drug Research and Development with Genomics

Jeong-Sun Seo, Seoul National University Bundang Hospital and Macrogen, South Korea
GenomeAsia 100K (GA100K) and Asian Reference Genome

Martin G. Reese, Fabric Genomics, USA

Accurate and Rapid WGS Interpretation – In Clinical Care

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

SATURDAY, JUNE 9

Departure