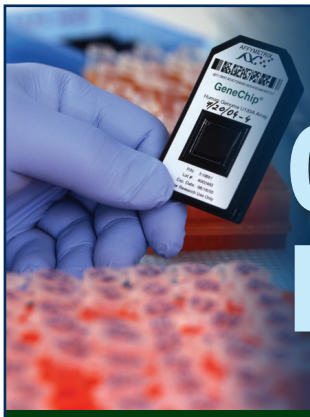

One Million Genomes: From Discovery to Health

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

KEYSTONE SYMPOSIA™
on Molecular and Cellular Biology
Accelerating Life Science Discovery

www.keystonesymposia.org/meetings | 1.800.253.0685 | 1.970.262.1230

a 501(c)(3) nonprofit educational organization

KEYSTONE SYMPOSIA
on Molecular and Cellular Biology

One Million Genomes: From Discovery to Health (G1)

June 4-8, 2018 • Herrenhausen Palace • Hannover, Germany

Scientific Organizers: Geoffrey S. Ginsburg, Teri Manolio and Patrick Boon Ooi Tan

Organized in collaboration with Volkswagen Foundation

Sponsored by Regeneron Pharmaceuticals, Inc. and Vertex Pharmaceuticals Incorporated

Global Health Travel Award Deadline: January 9, 2018 / Abstract & Scholarship Deadline: February 21, 2018 / Abstract Deadline: March 27, 2018 / Discounted Registration Deadline: April 11, 2018

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

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 Cencer Research Center, USA
Multiplex NGS Testing in Cancer Care: An Economic Perspective on Hope, Hype and Value

Sarah Wordsworth, 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