FINAL AGENDA

Register by March 29

SAVE up to $550!

Hotel Kabuki, San Francisco, CA
June 25 - 28, 2013

THE 2ND ANNUAL

TCGC CLINICAL GENOME CONFERENCE

Advances in Clinical Genome Sequencing, Diagnostics and Interpretation

PART ONE
June 25 - 26, 2013

TCGC: The SCIENCE of Investigation and Interpretation

PART TWO
June 27 - 28, 2013

TCGC: The BUSINESS of Integration and Implementation

KEYNOTE PRESENTATIONS

Integrating Genome Sequencing into Clinical Medicine: When, Why, and How?
Bruce R. Korf, University of Alabama at Birmingham

Delivering Genomic Medicine: Challenges and Opportunities
Heidi L. Rehm, Partners Healthcare Center for Personalized Genetic Medicine

Progress in Aggregating All the World's Genetic Tests into a Single Assay
Randy Scott, InVitae

Adventures in Personal Medicine: Integrated Personal Omics Profiling for Following Healthy and Disease States
Michael Snyder, Stanford University

Genetics of Common/Complex Clinical Problems
Kári Stefánsson, deCODE genetics

Regulatory Considerations and Challenges for Ultra-High-Throughput Sequencing-Based Clinical Applications
Živana Težak, FDA

Co-Located With:
TCEC The Clinical Epigenome CONFERENCE
June 26 - 27, 2013

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ClinicalGenomeConference.com

Conference-at-a-Glance*

Monday, June 24	Tuesday, June 25	Wednesday, June 26	Thursday, June 27	Friday, June 28
	TCGC: The Science of Investigation and Interpretation		TCGC: The Business of Integration and Implementation	
		TCEC: The Clinical Epigenome Conference		
Pre-Conference Dinner Short Courses**	Welcome Reception	Dinner Short Courses**	Reception	

*For the complete and up-to-date agenda, visit ClinicalGenomeConference.com

**Separate Registration Required

Pre-Conference Dinner Short Courses** Monday, June 24

2:00 - 5:00 pm

(SC1) Implementing Next-Generation Sequencing for Clinical Diagnostics

The rapid evolution of next-generation sequencing and the resulting move into routine clinical practice requires arguably as much skill in navigating bureaucracy as mapping and interpreting base pairs. Significant challenges for clinical diagnostics include the rapid evolution of platforms, protocols, kits, and reagents as well as genome analysis, interpretation and ethics. This short course provides practical information on implementing clinical sequencing, genomic data analysis and interpretation, ethics, CLIA certification and CAP standards. This course has been created in collaboration between Cambridge Healthtech Institute and the College of American Pathologists.

Instructors:

Development of Laboratory Standards for Next-Generation Sequencing as a Clinical Tool

Nazneen Aziz, Ph.D., Director of Molecular Medicine, Transformation Program Office, College of American Pathologists

Data Interpretation and Ethics in Clinical Genomic Sequencing

Wayne W. Grody, M.D., Ph.D., Professor, Medical Genetics and Molecular Diagnostics; Director, Clinical Genomics Center, School of Medicine, University of California, Los Angeles

5:30 - 8:30 pm

(SC2) Getting Personal About Genomes

Having your exome or whole genome sequenced is now feasible. However, the prospect of analyzing and interpreting your personal genome is not that simple. Getting Personal about Genomes is hosted by bioinformaticians who have the expertise, tools, and vested interest in analyzing and interpreting their own genomes and are willing to share their journeys. As these trailblazers shed light on their genomes, they are advancing the understanding of genomics for all.

Instructors:

Open-Source Platforms for Personal Genomics

Konrad J. Karczewski, Biomedical Informatics, Stanford University and Co-author, "Exploring Personal Genomics"

The Analysis and Interpretation of My DTC 23andMe Exome

Gabe Rudy, Vice President, Product Development, Golden Helix and Author, "A Hitchhiker's Guide to Next Generation Sequencing"

Dinner Short Courses** Wednesday, June 26 6:00 - 9:00 pm

(SC3) Clinical Combination Economic Conundrums

The explosion of molecular and imaging findings presents potentially economically disruptive choices with powerful outcome implications for clinicians, payers, product innovators and patients. We are discovering that the rapid-fire scientific advances are not always paced by clinical translation into products, medical community adoption and population health benefits. This short course will provide participants with an integrated understanding of the knowledge, funding, regulatory, legal and economic incentive gaps that are slowing our progress, as well as the potential impact of health care reform and the myriad of solutions being explored such as Silicon Valley open innovation, creative public/private collaborations, traditional industry and federal models to new approaches using tools such as adaptive licensing, coverage with evidence and "NASA-like" outcome-driven programs. Using this integrated framework, multiple innovative new paths to success emerge for clinicians, product innovators and patient advocates.

Instructor:

Mark Trusheim, Executive in Residence & Visiting Scientist at MIT; former Special Government Employee, Office of the Commissioner, FDA

(SC4) Advances in Methylation Analysis

The introduction of high-throughput whole-genome methylation analysis techniques has produced a startling amount of information revealing the accumulation and impact of aberrant methylation patterns across disease epigenomes – particularly within human cancers. More recently, with the inclusion of 5hmC analysis, the rapid expansion of tools create a plethora of options, leaving researchers to navigate multiple assays and analysis strategies. This workshop features a series of lectures from technology experts providing practical guidance for 5mC and 5hmC analysis for clinical applications.

Instructors:

Genome-Wide Bisulfite Sequencing Using NGS

Huidong Shi, Ph.D., Associate Professor, Biochemistry and Molecular Biology, Medical College of Georgia, Georgia Health Sciences University Cancer Center

Quantifying Active De-Methylation with TAB-Seq

Gary Hon, Ph.D., Senior Researcher, Ren Lab, Cellular and Molecular Medicine, Ludwig Institute for Cancer Research, University of California, San Diego

Genome-Wide Hydroxymethylation Using HELP-GT

Dr. Yiting Yu, Ph.D., Instructor, Department of Epidemiology and Population Health, Albert Einstein College of Medicine

Additional Instructors to be Announced

**Separate Registration Required

Part One:

TCGC: The Science of Investigation and Interpretation

TUESDAY, JUNE 25

7:45 am Conference Registration and Morning Coffee

Opening Keynote Session

8:30 Chairperson's Opening Remarks
Kevin Davies, Ph.D., Editor-in-Chief, Bio-IT World

8:45 Integrating Genome Sequencing into Clinical Medicine: When, Why, and How?
Bruce R. Korf, M.D., Ph.D., Wayne H. and Sara Crews Finley Chair in Medical Genetics; Professor and Chair, Department of Genetics; Director, Heflin Center for Genomic Sciences, University of Alabama at Birmingham
The technology of genome sequencing is advancing much more rapidly than the knowledge of how to use the large volume of genomic information to achieve better prevention and management of disease. This talk will review some of the possible clinical applications of genome sequencing, the barriers to implementation, and some approaches that may help to overcome these barriers.

9:30 Delivering Genomic Medicine: Challenges and Opportunities
Heidi L. Rehm, Ph.D., FACMG, Assistant Professor of Pathology, Brigham and Women's Hospital and Harvard Medical School; Director, Laboratory for Molecular Medicine, Partners Healthcare Center for Personalized Genetic Medicine
This talk will address our experience in offering genomic sequencing services, including data analysis and clinical interpretation and reporting. It will address innovative solutions to increase the efficiency of genomic reporting through software solutions and widespread data sharing.

10:15 Morning Coffee Break

10:30 Progress in Aggregating All the World's Genetic Tests into a Single Assay
Randy Scott, Ph.D., Chairman and CEO, InVitae
Rare genetic diseases have long suffered from a lack of research due to limited commercial interest. However, there are thousands of Mendelian inherited genetic disorders and millions of affected individuals worldwide. Our goal is to aggregate all of the world's genetic tests into a single assay for less than the cost of a single gene assay today by applying the power of next-generation DNA sequencing and allowing routine testing for even rare conditions.

11:15 Genetics of Common/Complex Clinical Problems
Kári Stefánsson, M.D., Dr. Med., CEO, President, and Director, deCODE genetics

12:00 pm Close of Session

12:15 Luncheon Presentation: Routine, Reliable Genomic Interpretation: A Call for a Fully Integrated System of Software and Reference Data
Sponsored by Knome
Ben Salisbury, Ph.D., Vice President, Clinical Genomics, Knome, Inc.
By its nature, genome sequencing uncovers millions of variants ranging from clinically understood to novel and ambiguous. To address this variety, an analysis pipeline must accurately map each variant to common coordinates and tie it together with disparate external sources of information and predictive assessments. Institutional knowledge must also be incorporated. In a clinical context, this process must be reliable, repeatable, controlled, and readily validated. A solution that satisfies many of these needs will be presented.

Sample Collection

1:45 Chairperson's Remarks

1:50 EngageUC: Advancing Biorepository Research at the University of California
Daniel Dohan, Ph.D., Associate Professor & Associate Director, Training and Development, Philip R. Lee Institute for Health Policy Studies, University of California, San Francisco
Elizabeth Boyd, Ph.D., Associate Vice Chancellor, Ethics and Compliance; Program Director, Regulatory Knowledge and Support Program, CTSI; Associate Adjunct Professor, Social and Behavioral Science, University of California, San Francisco
Sarah Dry, M.D., Co-Director, Translational Pathology Core Laboratory; Associate Professor, Pathology and Laboratory Medicine, David Geffen School of Medicine, University of California, Los Angeles
EngageUC is an NIH-funded project to advance biorespository research among 5 University of California medical centers that collectively serve approximately 12 million individuals across the state. The 3-year project will develop uniform biorepository operations and harmonized biorepository governance practices for sample and data handling, system-wide IRB and regulatory approvals, best practices to consent outpatients for remnant sample collection, and ongoing relationships with California's diverse communities via an innovative community engagement component.

2:25 The Era of Clinical Whole Genome Sequencing and Personalized Medicine
Michael Christman, Ph.D., President and CEO, Coriell Institute for Medical Research
Access to patient genomic information will become increasingly important as physicians are progressively more receptive to incorporating genomics into routine clinical practice. When a patient needs a new prescription, it will be necessary for the physician to quickly and securely access your genetic data to understand drug efficacy prior to dosing. Who will patients and medical professionals trust to store and interpret the data? Coriell and the CPMC research study have defined several of the key barriers to accelerate the adoption and routine use of genomics in medicine and proposed effective solutions that are generally applicable.

3:00 Afternoon Refreshment Break

Genome Interpretation Software Solutions

3:20 Software Spotlights *(Sponsorship Opportunity Available)*
Obtaining clinical genome data is rapidly becoming a reality but analyzing and interpreting the data continues to be a bottleneck. While there are many commercial software solutions and pipelines for managing raw genome sequence data, providing the medical interpretation and delivering a clinical diagnosis will be the critical step in making good on the promise of genomic medicine.

Interpretive Analytics of Big Data in Oncology by Physicians, for Physicians: Supporting Clinical Decision-Making at the Point of Care
Sponsored by CollabRx
Gavin J. Gordon, Ph.D., Head, Business Development & Alliances, CollabRx, Inc.

Turnkey Next-Generation Diagnostics
Sponsored by SVBio
Dietrich A. Stephan, Ph.D., CEO, SV Bio

Automated Genome-Phenome Analysis to Solve the Clinical Interpretation Bottleneck
Sponsored by SimulConsult
Michael Segal, M.D., Ph.D., Founder & Chief Scientist, SimulConsult

Towards Simplifying Genomic Medicine
Sponsored by Strand
Ramesh Hariharan, Ph.D., Founder & Chief Technologist, Strand Life Sciences

You Can't Interpret What You Don't See: Accurate and Fast Variant and *de novo* Mutation Identification from NGS Data
Sponsored by REAL TIME GENOMICS
Francisco M. De La Vega, D.Sc., Vice President, Genome Science, Real Time Genomics
Identifying variants from NGS data persistently results in false negatives and positives, confounding downstream analysis and interpretation. We developed a solution that leverages family relationships to fast and accurately identify variants and *de novo* mutations in trios, reducing false negatives. Exome data can be aligned and called in just hours.

5:05 Close of Session

5:15 Welcome Reception in the Exhibit Hall with Poster Viewing

6:30 Close of Day

WEDNESDAY, JUNE 26

7:30 am Breakfast Presentation *(Sponsorship Opportunity Available)* **or Morning Coffee**

Identifying Clinically Relevant Variants

8:30 Chairperson's Remarks

8:35 The Need for CLARITY in Clinical Genome Sequencing

Catherine A. Brownstein, Ph.D., MPH, Gene Partnership, Boston Children's Hospital; Department of Pediatrics, Harvard Medical School

The CLARITY competition was launched with the goal of surveying current practices in bioinformatic analysis, interpretation, and reporting of next-generation sequencing to diagnose rare genetic conditions. This international challenge provided contestants with DNA sequences and clinical data from three families with rare conditions for which no genetic cause had been identified. The presentation will summarize results and discuss some of the conclusions and lessons from this competition.

9:10 Identification of Disease Alleles from Whole Genome and Exome Sequencing Data Using a Unified Framework for Ranking Genetic Variation

Martin Kircher, Ph.D., Senior Fellow, Jay Shendure's Lab, University of Washington

One remaining challenge in the analysis of genetic data is the interpretation of genetic variation and the identification of the few phenotypically causal variants or disease variants among the few million variants present in each sequenced genome. While various programs for assessing the functional impact of variants exist, they are largely limited to highly conserved positions in protein-coding sequences. We present a unified approach that integrates diverse types of available annotations and scores into a single framework that weights the functional impact of both coding and non-coding variation on a genome-wide scale.

9:45 Challenges in Whole Genome Sequencing of Complex Traits: Analytical Pipelines and Data Integration for "Big" Genomics Data

Fabio Macciardi, M.D., Ph.D., Professor, Department of Psychiatry and Human Behavior; Director, Core Genomics, University of California, Irvine

The exceptional potential of the use of sequencing as a tool for the development of medicine will not be exploited if we do not deal appropriately with the complexity of phenotypes in complex traits. I will describe our approach to these complexities, briefly presenting the Graphical Pipeline for Computational Genomics (GPCG). The GPCG implements flexible workflows that cover all the analytical steps required for NGS data, using a distributed client-server infrastructure for computational analysis. As a working example of this integrated approach, we will then show an application to genomics data of schizophrenia, where the integration of DNA and RNA sequencing is potentially revealing unexpected mechanisms of disease.

10:20 Coffee Break in the Exhibit Hall with Poster Viewing

11:00 Optimizing Analysis Pipelines for Improved Variant Discovery from Personal Genomes

David Mittelman, Ph.D., Associate Professor, Virginia Bioinformatics Institute, Virginia Tech; Associate Professor, Department of Basic Science, Virginia Tech Carilion School of Medicine

There are many mapping strategies and variant calling methods, and while variant callers tend to identify similar base substitutions, they often differ substantially in assigning indels and copy number variants. It is therefore, critical to optimize all aspects of the analysis pipeline to maximize detection of variants, while minimizing the false discovery rate. As a case study we present our analysis pipeline for detecting human microsatellite repeat variation. Repeats are challenging to genotype, however they are an important source of clinical and subclinical variation.

11:35 PANEL DISCUSSION: GCAT: Genome Comparison & Analytic Testing - Uniting on Standards through Crowdsourcing

Panel Moderator: David Mittelman, Virginia Tech Carilion School of Medicine

The standardization and performance testing of genome analysis tools is a prerequisite to widespread adoption of high-throughput sequencing, particularly in the clinic. We present an open and collaborative platform for comparing multiple genome analysis tools across a standard set of metrics. The exact metrics and datasets are crowdsourced to encourage community involvement and input. The GCAT platform features an easy interface and automatically generates compelling visualizations of benchmark and performance testing data.

Panelists to be Announced

12:10 pm Close of Session

12:15 Luncheon Presentation *(Sponsorship Opportunity Available)* **or Lunch on Your Own**

Identifying Clinically Relevant Variants cont.

1:30 Chairperson's Remarks

1:35 Talk Title to be Announced

Hanlee Ji, M.D., Assistant Professor, Division of Oncology, Department of Medicine, Stanford University School of Medicine; Senior Associate Director, Stanford Genome Technology Center

2:10 The Clinical Exome and Symptom-Guided Analysis

Elaine Lyon, Ph.D., Associate Professor of Pathology, University of Utah School of Medicine; Medical Director of Molecular Genetics/Genomics, ARUP Laboratories

As next-generation sequencing technologies improve in accuracy and cost effectiveness, they are being integrated into clinical diagnostics. However, interpretation of the exome or genome remains a challenge. This presentation will review analysis steps to identify clinically significant variants. Strategies to prioritize relevant variants based on a patient's symptoms will be discussed.

2:45 Streamlining Genetic Diagnostics for Patients with Consanguinity

Anna Lehman, M.D., FRCPC, Assistant Professor, Medical Genetics, Adult Metabolic Diseases, University of British Columbia

Patients with consanguineous ancestry presenting with congenital diseases have a high likelihood of having an autosomal recessive genetic cause. If a single gene candidate is not clinically recognizable, then SNP microarray is usually the best next step. Perusal of OMIM diseases within large regions of homozygosity may indicate the likely causative gene. If that step fails, then exome sequencing, ideally in a different affected relative if available, will provide a short list of homozygous rare damaging variants within the regions of homozygosity. In the presenter's experience using this parsimonious protocol with consecutive, unselected patients, the diagnostic yield exceeds 50%.

3:20 Toward More Accurate Variant Calling for "Personal Genomes"

Gholson Lyon, M.D., Ph.D., Assistant Professor, Human Genetics, Cold Spring Harbor Laboratory

We are working to optimize variant calling in personal genomes, including SNVs and indels, and we are leveraging the power of families to increase accuracy. There is still substantial room for improvement in attaining a clinical-grade comprehensive set of variants with high sensitivity and specificity in any one personal genome. Further ancestry tracking and family history data, when combined with whole genome sequencing, can facilitate genetic prediction for rare, highly penetrant diseases in families.

3:55 Refreshment Break in the Exhibit Hall with Poster Viewing

» 4:30 PLENARY KEYNOTE PRESENTATION

Adventures in Personal Medicine: Integrated Personal Omics Profiling for Following Healthy and Disease States

Michael Snyder, Ph.D., Professor and Chair, Genetics; Director, Stanford Center for Genomics and Personalized Medicine, Stanford University

Personalized medicine is expected to benefit from the combination of a person's DNA sequence with a global and detailed molecular monitoring whereby thousands of molecules are followed to assess physiological states. To ascertain whether this can be achieved, we have been sequencing the whole genome sequence of individuals to estimate disease risk and follow 40,000 molecules in the blood. This analysis results in an integrated Personal Omics Profile (iPOP) that reveals extensive, dynamic and broad changes in diverse molecular components and biological pathways across healthy and disease conditions. This analysis was applied to an individual and was used to predict Type 2 Diabetes, whose onset was observed during the course of our study. Our work is expected to be important for performing personalized medicine.

5:15 Close of TCGC: The Science of Investigation and Interpretation

5:30 Registration for Dinner Short Course and TCGC: The Business of Integration and Implementation

6:00 - 9:00 Dinner Short Courses *(See page 2 for details)*

Part Two:

TCGC: The Business of Integration and Implementation

THURSDAY, JUNE 27

7:30 am Conference Registration and Morning Coffee

Policy, Privacy and Patient Care

8:30 Chairperson's Opening Remarks

» 8:40 KEYNOTE PRESENTATION

Regulatory Considerations and Challenges for Ultra-High-Throughput Sequencing-Based Clinical Applications

Živana Težak, Ph.D., Associate Director, Science and Technology, Personalized Medicine, Office of In Vitro Diagnostic Device Evaluation and Safety (OIVD), Center for Devices and Radiological Health (CDRH), FDA

The use of diagnostic tests that determine genetic sequence variation to guide patient care is rapidly expanding. At the same time, this rapid development of genomic sequencing technology and applications is raising new policy and regulatory questions. Some of the questions include efficient approaches for adequate demonstration of analytical performance. While there are no broadly accepted standards or methods to analytically validate ultra-high-throughput sequencing tests yet, there are a number of efforts underway, including establishing reference material that can aid in assessing analytical performance. The additional challenge in informed clinical application of sequencing information is inadequate knowledge about correlations between genomic variations and their clinical significance. Although there is a wealth of data being generated about genetic variation, a lot of it is fragmented and there is a need for concerted efforts to curate and organize this knowledge in a way that can support its clinical use. FDA is taking a number of steps to encourage cross-collaboration in this area, to be able to balance public safety concerns with fostering innovation and enabling the translation of emerging technologies that can improve medical care and benefit public health.

9:25 The Current State of Gene Patents

John Conley, J.D., Ph.D., William Rand Kenan, Jr. Professor of Law, University of North Carolina

This presentation will analyze the current state and likely future of gene patents. A primary focus will be on the ongoing AMP vs. Myriad Genetics case, now before the Supreme Court, which is the first case to raise squarely the question of whether and in what forms DNA sequences can be patented. A decision is possible before the conference, so the presentation may be able to provide a timely report on what the Court has decided. The presentation will also review other, less publicized ways in which gene patents have been effectively challenged, including through the nonobviousness and written description requirements. Moreover, current and future sequencing technologies may be bypassing single-gene patent claims, rendering them less valuable regardless of what the Supreme Court decides, and motivating companies to seek other kinds of commercial advantages. Myriad Genetics' announced business strategy for Europe will be discussed as a reflection of this new reality.

10:00 Sponsored Presentation *(Opportunity Available)*

10:15 Coffee Break in the Exhibit Hall with Poster Viewing

11:00 Privacy and Progress in Whole Genome Sequencing: The Presidential Commission Report

Elizabeth Pike, J.D., LL.M., Senior Policy and Research Analyst, Presidential Commission for the Study of Bioethical Issues

In October 2012, the Presidential Commission for the Study of Bioethical Issues released its report Privacy and Progress in Whole Genome Sequencing. The report concludes that to realize the enormous promise that whole genome sequencing holds for advancing clinical care and the greater public good, individual interests in privacy must be respected and secured. The Commission offers 12 recommendations to help craft policies that are flexible enough to ensure progress and responsive enough to protect privacy. In this session, we will discuss some of the ethical and privacy concerns raised by the increased use of whole genome sequencing and the ways that the Commission chose to address these concerns.

11:35 Mapping the Next-Generation Sequencing Industry

Margaret Curnutte, Ph.D., Research Fellow, Baylor College of Medicine

This presentation provides an overview of the emerging business models within the next-generation sequencing industry. From sequencing to variant calling to annotating and analyzing and data storage, companies in this area of innovation are developing and/or pairing such features in novel ways that challenge policy implications for the clinic. These implications will be discussed with reference to the current regulatory landscape for genetic testing technologies.

12:10 pm Close of Session

12:15 Luncheon Presentation *(Sponsorship Opportunity Available)* **or Lunch on Your Own**

1:30 Chairperson's Remarks

The Clinical Genome Technology Showcase *(Sponsorship Opportunities Available)*

The success of producing As, Cs, Gs, and Ts at exponentially lower costs, along with longer and more accurate reads, is revolutionizing genomic diagnostic medicine. Thus, the promise of next-generation sequencing has shifted from discovery to clinical utility. Now the question is: how do we best use this sequence information in patient care? Hear first-hand from the companies that continue to drive the genomics revolution.

1:35 Clinical Interpretation of Genomes in Healthy Individuals

Sponsored by illumina

Tina Hambuch, Ph.D., Staff Scientist, Illumina

2:05 - 3:35 Additional Sponsored Presentations

Sponsored by N of One

3:05 - 3:20 Interpreting Molecular Data from Sequencing Platforms with Leading Edge Knowledge for Treatment and Diagnostic Options

Jennifer Carter, Founder & CMO, N-of-One, Inc.

Physicians, labs, institutions and patients can efficiently make the best diagnostic and treatment choices, The PrecisionWorks framework enables experts to translate molecular data specific to each patient and the extensive available knowledge for each cancer type into state-of-the-art, clinically actionable insights and therapeutic options focused at the point of care.

3:35 Refreshment Break in the Exhibit Hall with Poster Viewing

Genomic Data Integration

4:15 The New NIH Genetic Testing Registry: Bringing Together Science, Medicine and Policy

Wendy Rubinstein, M.D., Ph.D., Senior Scientist and Director, NIH Genetic Testing Registry, National Center for Biotechnology Information, National Library of Medicine, National Institutes of Health

Dr. Rubinstein will introduce the GTR, a free online resource that provides centralized access to comprehensive genetic test information that is voluntarily submitted by test providers. She will survey the content of GTR including registered laboratories, accessioned tests for heritable conditions, and the quality measures and evidentiary basis of the tests. NCBI's representation of information about clinically relevant variants will also be discussed.

4:45 Galaxy in a Clinical Setting: Architecture and Storage Implications

Sponsored by EMC® ISILON

Sanjay Joshi, CTO, Life Sciences, EMC Isilon Storage Division

James Taylor, Assistant Professor, Biology, Emory University

The need for a process-level source and version control, configuration management and document life-cycle management system is reaching critical path as Research Genomics translates into Clinical Genomics. We will discuss the architectural and storage requirements for using Galaxy, the process management platform of choice for Genomics, in a Clinical environment.

5:15 Evening Reception

6:30 Close of Day

FRIDAY, JUNE 28

7:30 am Breakfast Presentation *(Sponsorship Opportunity Available)* **or Morning Coffee**

Dynamics of Genomic Data Implementation

8:30 Chairperson's Remarks

8:35 Advanced Sequencing; Finding the Path from Research to the Clinical Lab in a Dynamic Landscape

Jamie L. Platt, Ph.D., CGMBS, MB(ASCP), Scientific Director, Advanced Sequencing, Quest Diagnostics Nichols Institute

Advances in sequencing technology bring promises of a new era of genomic and individualized medicine. However, the clinical lab setting is very different from the research environment and warrants specific considerations around workflow, standardization, pipeline, costs, and regulatory issues, among others. The considerations for moving advanced sequencing into the clinical setting will be discussed.

9:10 Development and Implementation of NGS Antiviral Drug Resistance Assays in a Clinical Reference Laboratory Environment: Progress and New Challenges

Christos Petropoulos, Ph.D., VP, R&D; CSO, Monogram Biosciences (a LabCorp Company)

I will discuss the plans, progress and specific technical and analytical challenges related to developing, validating and implementing NGS assays in a clinical reference laboratory environment. The presentation will focus on the development of antiviral drug resistance assays that are being used to support the pre-clinical and clinical evaluations of new drug candidates for the treatment of HIV and HCV infections. Many such assays are expected to move into patient treatment settings in the clinic and at least one assay has already made this transition. I will also provide an broad overview of other NGS efforts at LabCorp, including the establishment of prognostic and diagnostic assays for oncology and genetics applications.

9:45 Clinical Grade Gene Expression Signature Development Using RNA-seq

Vinay Varadan, Ph.D., Senior Member Research Staff, Clinical Bioinformatics, Philips Research North America

RNA-sequencing has the potential to accelerate the process of translating discovered gene expression signatures into clinical grade assays. However, developing a clinically applicable assay requires standardization at every step, from tissue handling until final signature readout, and is fraught with pitfalls that need to be systematically addressed to achieve clinical-grade utility. In this talk, we will outline our experience in analyzing breast tumor tissues from a multi-site clinical trial using RNA-seq. We were able to recapitulate existing gene expression tests such as ER/PR/HER2 and the PAM50 subtyping signature using RNA-seq and in addition discover a TGFb signature predictive of clinical benefit to neoadjuvant-bevacizumab. Despite the fact that the tumor samples in our study were processed at multiple sites and the sequencing performed on different Illumina sequencers, we were able to show technical validation of the signature using gold-standard RT-PCR technology. We will outline the steps we are taking to develop a clinical-grade assay of the discovered signature and identify the bottlenecks and challenges in creating an RNA-seq-based clinical assay.

10:20 Sponsored Presentation *(Opportunity Available)*

10:35 Coffee Break, Last Chance for Exhibit & Poster Viewing

11:15 PANEL DISCUSSION: Payer's Dilemma – Panels, Exomes, or Whole Genomes?

Panel Moderator: Katherine Tynan, Ph.D., Business Development & Strategic Consulting for Diagnostics Companies, Tynan Consulting LLC

As the cost of clinical exome and genome sequencing falls, the use of this technology to end diagnostic odysseys is becoming more widespread. This raises a host of perplexing questions, including whether insurance companies will pay for the procedure. In a best-case scenario, sequencing a patient's full genome can lead to a precise diagnosis and inform a new therapeutic strategy; moreover, the cost of WGS is now comparable to many single-gene tests or panels. On the other hand, sequencing an entire genome has not been proven beneficial in any clinical trials, and raises additional complexities such as the fate of incidental/secondary data. There's also the question of whether CPT codes are moving quickly enough to embrace the new technology.

In this panel discussion, experts will discuss the impact of new clinical genomics technologies on reimbursement.

Panelists to be Announced

12:00 pm Luncheon Presentation *(Sponsorship Opportunity Available)* **or Lunch on Your Own**

Dynamics of Genomic Data Implementation cont.

1:30 Chairperson's Remarks

1:35 The Coverage Conundrum: Unmet Need and Cutting-Edge Science Don't Necessarily Translate into Patient Access

Pam Baker, Senior Director, Market Access & Policy, CardioDx, Inc.

Advances in genetics and genomics have the potential to make a significant impact on patients' lives, but great science paired with a large market opportunity are only part of the story. Without payer coverage for most of these important advancements, many will end up as interesting concepts that weren't able to impact the practice of medicine in any meaningful way. We'll discuss how to navigate the sometimes unclear requirements for coverage by payers both public and private and what those requirements mean for the clinical development plan early on. We'll also share some of the main stumbling blocks to payer coverage for diagnostics, why they exist, and recommendations on how to avoid them.

2:10 Talk Title to be Announced

Gary Palmer, M.D., Senior Vice President, Medical Affairs, Foundation Medicine, Inc.

2:45 Talk Title to be Announced

Richard Boles, M.D., Medical Director, Courtagen

3:35 PANEL DISCUSSION: Diagnostics' Dilemma – Panels, Exomes, or Whole Genomes?

The world of molecular diagnostics is undergoing a revolution as whole-exome and -genome sequencing enters the clinic. For years, diagnostics firms have offered single-gene tests (e.g. BRCA1, CFTR) or gene panels (e.g. ataxia, neuropathy). New technologies threaten to disrupt this lucrative business by casting a far bigger net – providing a "super panel" for the entire exome or whole genome. What are the pros and cons of this approach? Will the molecular diagnostics industry embrace change?

In this panel discussion experts from all sides will debate the impact of new genomic technologies as they march into the clinic.

Panelists

Andrew Grupe, Ph.D., Senior Director Pharmacogenomics, Celera/Quest Diagnostics

4:30 Close of TCGC: The Business of Integration and Implementation

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CHI offers comprehensive sponsorship packages which include presentation opportunities, exhibit space and branding, as well as the use of the pre and post-show delegate lists. Customizable sponsorship packages allow you to achieve your objectives before, during, and long after the event. Signing on early will allow you to maximize exposure to hard-to-reach decision makers!

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To customize your participation at this event, please contact:
Jay Mulhern – Business Development Manager
781-972-1359 | jmulhern@healthtech.com

HOTEL & TRAVEL INFORMATION

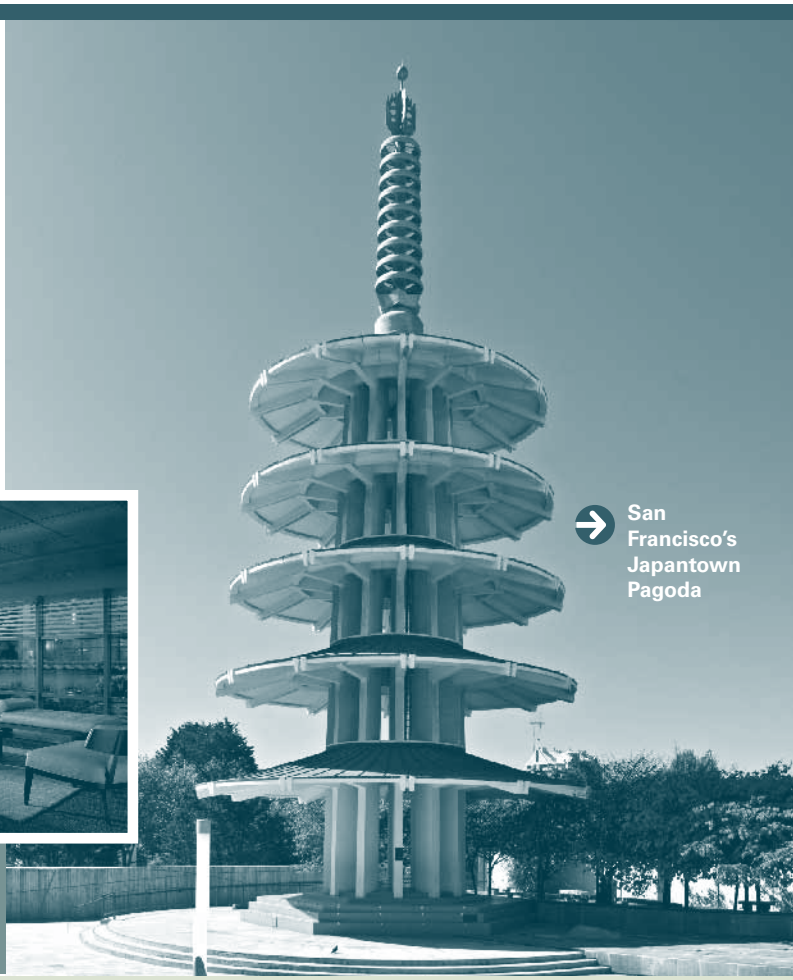
Conference Hotel:
Hotel Kabuki
1625 Post Street
San Francisco
Phone: 415-922-3200

Discounted Room Rate: $154 s/d
Discounted Room Rate Cut-off Date: May 27, 2013

Please visit our conference website to make your reservations online or call the hotel directly to reserve your sleeping accommodations. You will need to identify yourself as a Cambridge Healthtech Institute conference attendee to receive the discounted room rate with the host hotel. Reservations made after the cut-off date or after the group room block has been filled (whichever comes first) will be accepted on a space- and rate-availability basis. Rooms are limited, so please book early.

Top reasons to stay at THE HOTEL KABUKI

- Take advantage of discounted TCGC rates – only $154 per night!
- No Commute! Meeting takes place at the hotel.
- Adjacent to Japan Center - a large Japanese-themed shopping mall, boasting many stores, activities, and restaurants
- Less than 2 blocks away from restaurants & shopping on Fillmore Street, an energized & hip shopping district
- Explore & experience Japantown in the heart of the city



San Francisco's Japantown Pagoda

Pricing and Registration Information

SHORT COURSES

	Commercial	Academic, Government, Hospital-affiliated
One short course	$695	$395
Two short courses	$995	$595
Three short courses	$1195	$695

DINNER SHORT COURSE SELECTIONS	
Monday, June 24, 2:00 - 5:00 pm	Wednesday, June 26, 6:00 - 9:00 pm (Concurrent Courses)
(SC1) Implementing Next-Generation Sequencing for Clinical Diagnostics	(SC3) Clinical Combination Economic Conundrums
Monday, June 24, 5:30 - 8:30 pm	(SC4) Advances in Methylation Analysis
(SC2) Getting Personal about Genomes	

CONFERENCE PRICING

BASIC PACKAGE *(Includes access to 1 conference, excludes short courses)*

Early Registration Discount until March 29, 2013	$1495	$745
Advance Registration Discount until May 17, 2013	$1655	$825
Registrations after May 17, 2013, and on-site	$1865	$945

CONFERENCE SELECTIONS		
June 25 - 26 (Tuesday - Wednesday) **Part I: Science** of Investigation and Interpretation		June 27 - 28 (Thursday - Friday) **Part II: Business** of Integration and Implementation
	June 26 - 27 (Wednesday - Thursday) **The Clinical Epigenome Conference**	

Special Package Pricing: BEST VALUE! *(Includes access to the back-to-back Part I: Science of Investigation and Interpretation and Part II: Business of Integration and Implementation, as well as The Clinical Epigenome Conference, excludes short courses)*

Early Registration Discount until March 29, 2013	$2245	$1045
Advance Registration Discount until May 17, 2013	$2595	$1145
Registrations after May 17, 2013, and on-site	$2795	$1245

CONFERENCE DISCOUNTS

POSTER DISCOUNT ($50 Off) Poster abstracts are due by May 21, 2013. Once your registration has been fully processed, we will send an email containing a unique link allowing you to submit your poster abstract. If you do not receive your link within 5 business days, please contact jring@healthtech.com. *CHI reserves the right to publish your poster title and abstract in various marketing materials and products.*

ALUMNI DISCOUNT* (SAVE 20%): Cambridge Healthtech Institute (CHI) appreciates your past participation at Drug Discovery Chemistry. As a result of the great loyalty you have shown us, we are pleased to extend to you the exclusive opportunity to save an additional 20% off the registration rate.

NGS LEADERS DISCOUNT* (SAVE $50): Members of the NGS Leaders community will receive a $50 discount off the conference registration.

REGISTER 3 - 4th IS FREE*: Individuals must register for the same conference or conference combination and submit completed registration form together for discount to apply.

GROUP DISCOUNTS*: Discounts are available for multiple attendees from the same organization. For more information on group rates contact David Cunningham at +1-781-972-5472.

*Alumni, NGS Leaders, Twitter, LinkedIn, Facebook or any other promotional discounts cannot be combined. Discounts not applicable on event short courses.

If you are unable to attend but would like to purchase the TCGC: The Clinical Genome Conference CD for $500 (plus shipping), please visit ClinicalGenomeConference.com. Massachusetts delivery will include sales tax.

ADDITIONAL REGISTRATION DETAILS

Each registration includes all conference sessions, posters and exhibits, food functions, and access to the conference proceedings link.

Handicapped Equal Access: In accordance with the ADA, Cambridge Healthtech Institute is pleased to arrange special accommodations for attendees with special needs. All requests for such assistance must be submitted in writing to CHI at least 30 days prior to the start of the meeting.

To view our Substitutions/ Cancellations Policy, go to http://www.healthtech.com/regdetails

Video and or audio recording of any kind is prohibited onsite at all CHI events.

How to Register: **ClinicalGenomeConference.com**

reg@healthtech.com • P: 781.972.5400 or Toll-free in the U.S. 888.999.6288

Please use keycode **TCGC F** when registering!

Please refer to the Registration Code below:

Cambridge Healthtech Institute
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Needham, MA 02494
www.healthtech.com
Fax: 781-972-5425