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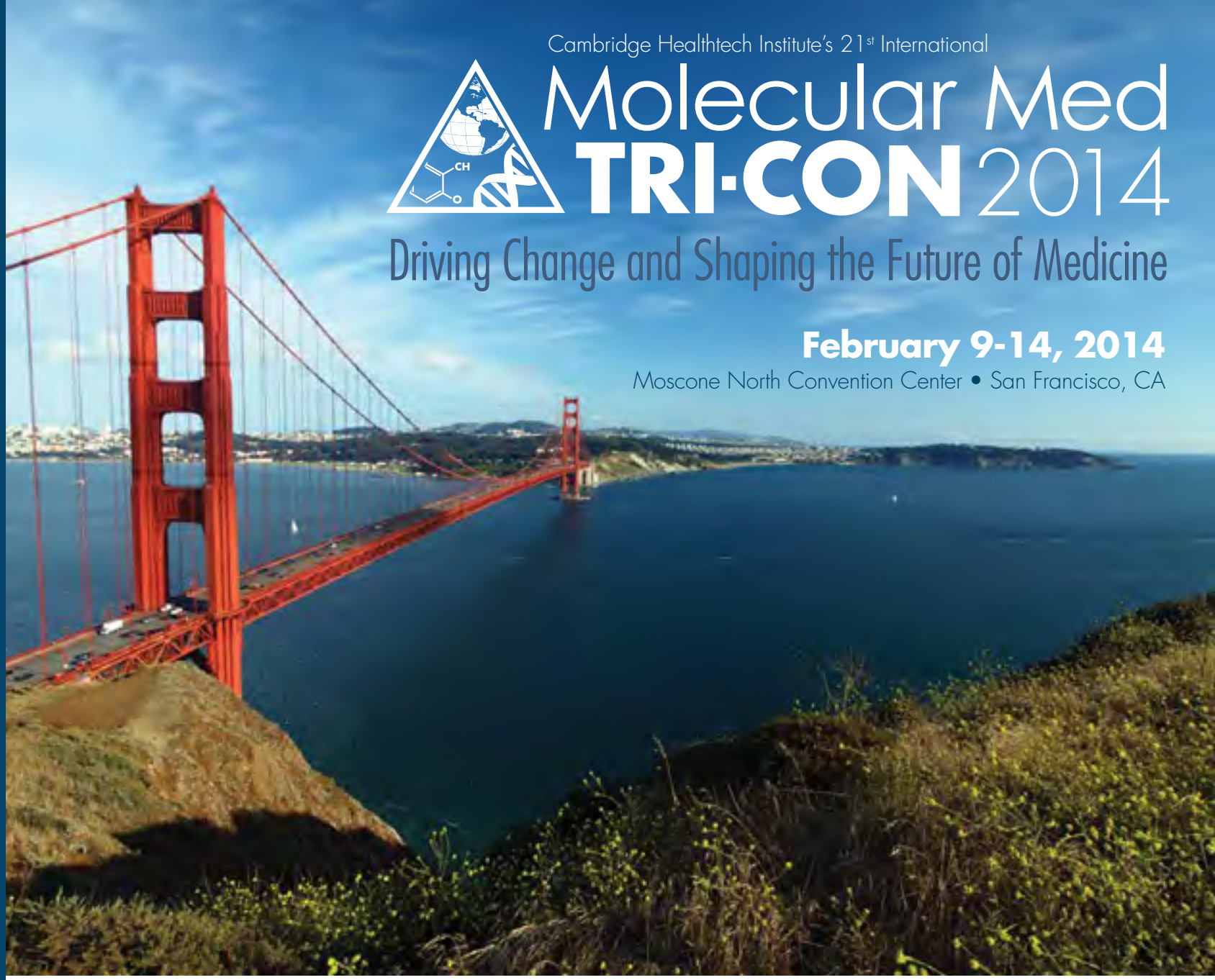

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February 9-14, 2014

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2014 EVENT-AT-A-GLANCE

EVENTS AT THE MOSCONE NORTH CONVENTION CENTER

Sunday, February 9

1:00 pm Registration

2:00 – 5:00 pm Afternoon Short Courses

5:30 – 8:30 pm Dinner Short Courses

Monday, February 10

7:30 am Registration and Morning Coffee

8:30 – 11:30 am Morning Short Courses

11:50 – 1:15 pm Conference Programs

1:15 – 2:30 pm Luncheon Presentations or Lunch on Your Own

2:30 – 5:00 pm Conference Programs

5:00 pm Plenary Keynote Session

6:15 – 7:45 pm Grand Opening Reception in the Exhibit Hall with Poster Viewing

Tuesday, February 11

7:00 am Registration and Morning Coffee

8:00 – 9:15 am Plenary Keynote Session

9:15 – 10:25 am Refreshment Break in the Exhibit Hall with Poster Viewing

10:25 am – 12:40 pm Conference Programs

12:40 – 1:40 pm Luncheon Presentations or Lunch on Your Own

1:40 – 2:15 pm Refreshment Break in the Exhibit Hall with Poster Viewing

2:15 – 4:20 pm Conference Programs

4:20 – 5:20 pm Valentine's Day Celebration in the Exhibit Hall

5:20 – 6:20 pm Breakout Discussions in the Exhibit Hall

Wednesday, February 12

7:00 am Registration and Morning Coffee

7:00 am Breakfast Presentations

8:00 – 9:45 am Plenary Keynote Session

9:45 – 10:35 am Refreshment Break & Poster Competition Winner Announced in the Exhibit Hall

10:35 am – 12:10 pm Conference Programs

12:10 – 1:00 pm Luncheon Presentations or Lunch on Your Own

1:00 – 1:40 pm Refreshment Break in the Exhibit Hall with Poster Viewing

1:40 – 5:35 pm Conference Programs

EVENTS AT THE WESTIN ST. FRANCIS HOTEL

Thursday, February 13

7:30 am Registration and Morning Coffee

9:00 am – 5:25 pm Symposia

Friday, February 14

8:00 am Morning Coffee

8:25 am – 12:30 pm Symposia

STAY CONNECTED

CONFERENCE PROGRAMS

DIAGNOSTICS CHANNEL

Molecular Diagnostics
Personalized Diagnostics
Cancer Molecular Markers
Circulating Tumor Cells
Digital Pathology
Companion Diagnostics
PCR for Molecular Medicine—NEW
Biospecimen Science and Sample Prep—NEW
Clinical Epigenetics—NEW
Genome and Transcriptome Analysis—NEW

CLINICAL CHANNEL

Clinical and Translational Science
Clinical Sequencing
Clinical Epigenetics—NEW

CANCER CHANNEL

Circulating Tumor Cells
Predictive Preclinical Models in Oncology
Cancer Molecular Markers
Clinical Epigenetics—NEW

INFORMATICS CHANNEL

Bioinformatics for Big Data
Integrated R&D Informatics and Knowledge Management
Genome and Transcriptome Analysis—NEW

SYMPOSIA

Targeting Cancer Stem Cells
Point-of-Care Diagnostics
Genomics in Medicine
Leaders in Precision Medicine—NEW
Circulating Cell-Free DNA—NEW
Genomics & Sequencing Data Integration, Analysis and Visualization—NEW

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Plenary Keynote Presentations

MONDAY, FEBRUARY 10

Keynote Introduction: *Alan Wu, M.D., Chief, Clinical Chemistry and Toxicology, San Francisco General Hospital; Professor, Laboratory Medicine, University of California, San Francisco*

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David R. Gandara, M.D.

Director, Thoracic Oncology Program; Professor and Senior Advisor to Director, UC Davis Cancer Center

David R. Gandara, M.D., Professor of Medicine at the University of California at Davis (UC Davis) School of Medicine, is the Senior Advisor to the Director, and Director of the Thoracic Oncology Program at UC Davis Comprehensive Cancer Center. He is a diplomat of the American Board of Internal Medicine specializing in Medical Oncology. He is currently principal investigator for a number of research projects in lung cancer, pharmacology trials at various phases, and Southwest Oncology Group (SWOG) trials. He is the principal investigator for a National Cancer Institute award to the California Cancer Consortium for Early Therapeutic Trials of New Anti-Cancer Agents. He is chair of the SWOG Lung Committee, and a member and prior co-chair of the NCI-directed Investigational Drug Steering Committee (IDSC). He has written over 300 articles, book chapters, abstracts and editorials. He is editor-in-chief of the journal Clinical Lung Cancer, serves on the editorial board of four oncology journals, and is a manuscript reviewer for eight additional journals. He served as president of International Association for the Study of Lung Cancer (IASLC) from 2009 to 2011 and is on the board of directors of IASLC and is a prior board member and secretary-treasurer of the American Society for Clinical Oncology (ASCO). He also is chair of the NCI-directed Lung Correlative Science Committee. After receiving his medical degree from the University of Texas Medical Branch in Galveston, Dr. Gandara was an intern and resident at the Madigan Army Medical Center in Tacoma and a fellow at the Letterman Army Medical Center Presidio of San Francisco.

TUESDAY, FEBRUARY 11

Keynote Introduction: *Frank White III, Ph.D., Director, Biology, Product Marketing, Elsevier*

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Kimberly L. Blackwell, M.D.

Professor of Medicine, Assistant Professor of Radiation Oncology; Director, Breast Cancer Program, Duke Cancer Institute

Dr. Blackwell is the Director of the Breast Cancer Program in the Duke Cancer Institute, where she oversees all basic and translational research programs involving breast cancer patients. She has played a major role in two recently approved breast cancer drugs, lapatinib and T-DM1, both of which were studied in her laboratory and developed in trials in which she served as principal investigator. Because of her work on promising new therapies that target tumor cells selectively, she was named one of TIME magazine's 100 most influential people in the world in 2013. For the past three years, she has served on the national Scientific Advisory Board of the Susan G. Komen Race for the Cure. She has reviewed for several grant committees (Department of Defense, Komen for the Cure) and peer-reviewed journals, including Journal of Clinical Oncology, Cancer Research, Clinical Cancer Research, and Cancer. She received her undergraduate degree at Duke University in Bioethics, and her medical degree at Mayo Clinic Medical School. Dr. Blackwell completed an internal medicine internship and residency and a hematology-oncology fellowship at Duke University Medical School. Dr. Blackwell has clinical and research interests in breast cancer angiogenesis, breast cancer in younger women, endocrine therapy, and HER2 targeted therapy for breast cancer. She maintains an active clinical practice focused on young women with breast cancer and has served as the PI or co-PI on more than 50 clinical trials in breast cancer. She is a past recipient of the Duke University Specialized Program of Research Excellence (SPORE) in breast cancer Young Investigator Award, the Duke Cancer Center Malek Family Award for outstanding cancer investigation, and the Joseph Greenfield Award for Research Mentorship. Dr. Blackwell has authored or co-authored more than 60 articles or book chapters appearing in journals such as Clinical Cancer Research, the Journal of Clinical Oncology, Cancer, Radiation Research, and Molecular Cancer Therapeutics.

C. Thomas Caskey, M.D.

FACP, FRSC, Professor, Department of Molecular and Human Genetics, Baylor College of Medicine

Dr. Caskey was the Director and CEO of the Brown Foundation Institute of Molecular Medicine at the University of Texas Health Science Center in Houston. Dr. Caskey previously served as Senior Vice President, Human Genetics and Vaccines Discovery at Merck Research Laboratories, West Point and as President of the Merck Genome Research Institute. Dr. Caskey is Board Certified in Internal Medicine, Medical Genetics, and Molecular Genetics with 25 years of patient care experience in these specialties. He is a member of the National Academy of Sciences, Institute of Medicine, Royal Society of Canada, member & Chair of the Board on Health Sciences Policy - Institute of Medicine and past President of the American Society of Human Genetics and the Human Genome Organization, and Texas Academy of Medicine, Engineering and Science. He is an editor of the Annual Reviews of Medicine. Dr. Caskey has received numerous academic and industry honors. His genetic research has identified the genetic basis of 10 major inheritable diseases and opened up the understanding of triplet repeat diseases (Fragile X, myotonic dystrophy and others). His personal identification patent is the basis of worldwide application for forensic science and he is also a consultant to the FBI in forensic science. His current research is focused on the genetic basis of schizophrenia. Dr. Caskey received his B.S. from the University of South Carolina, his M.D. from Duke University Medical School, and an honorary degree in Chemistry from the University of South Carolina.

WEDNESDAY, FEBRUARY 12

Plenary Keynote Panel:

Emerging Technologies and Industry Perspectives

Moderator: Michael H. A. Roehrl, M.D., Ph.D., Director, UHN Program in BioSpecimen Sciences, University of Toronto; Scientist, Ontario Cancer Institute

Panelists: Bernard F. Andruss, Ph.D., Vice President, CDx Development & Regulatory Affairs, Asuragen, Inc.
Rudi Pauwels, Ph.D., Executive Chairman & CEO, Biocartis
Yan Yang, Ph.D., Director, Lab Operations, in vivo Services, The Jackson Laboratory
Jeremy Bridge-Cook, Ph.D., Senior Vice President, Research & Development, Luminex
David C. Duffy, Ph.D., CTO, Quanterix Corporation
Gary Kennedy, CEO, Remedy Informatics

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SHORT COURSES*

AFTERNOON

SUNDAY, FEBRUARY 9 | 2:00 – 5:00 PM

CTCs from Bench to Bed: Streamlining from Research to Clinical Practice
Instructors: Marek Malecki, M.D., Ph.D., President, Phoenix Biomolecular Engineering Foundation; Visiting Professor, University of Wisconsin, Madison
Avraham Rasooly, Ph.D., Program Director, NCI NIH
John P. Langmore, Ph.D., CSO, Rubicon Genomics, Inc.

Latest Advances in Molecular Pathology
Co-Organized with cap
Instructors: Iris Schrijver, M.D., Professor of Pathology and Pediatrics; Director, Molecular Pathology Laboratory, Stanford University Medical Center, Lucile Packard Children's Hospital
James Zehnder, M.D., Associate Professor of Pathology and Medicine (Hematology), Stanford University Medical Center
Athena Cherry, Ph.D., Director, Cytogenetics Laboratory, Professor, Pathology and Pediatrics, Stanford Comprehensive Cancer Center

Starting an NGS Lab Part I: Technical Considerations
Co-Organized with AMP Association for Molecular Pathology
Instructors: Madhuri Hegde, Ph.D., FACMG, Associate Professor, Human Genetics; Executive Director, Emory Genetics Laboratory, Emory University School of Medicine
Monica J. Basehore, Ph.D., FACMG, Director, Molecular Diagnostic Laboratory, Greenwood Genetic Center
Birgit H. Funke, Ph.D., FACMG, Assistant Professor, Pathology, MGH/Harvard Medical School; Director, Clinical Research and Development, Laboratory for Molecular Medicine

Sequencing 101
Instructor: Ryan Kim, Ph.D., Director, UC Davis Genome Center, DNA Technologies and Expression Analysis Cores, University of California, Davis

Best Practices in Personalized and Translational Medicine
Instructors: Erik Bierwagen, Ph.D., Principal Programmer Analyst, Department of Bioinformatics, Genentech, Inc.
Doug Garrett, Senior Programmer Analyst in Research, Genentech
Brenda Yanak, Ph.D., Director, Precision Medicine Leader, Clinical Innovation, Pfizer

PCR Part I: qPCR and High Resolution Melting in Molecular Diagnostics
Instructors: Fred Russell Kramer, Ph.D., Professor, Department of Microbiology and Molecular Genetics, Public Health Research Institute, New Jersey Medical School
Carl Wittwer, M.D., Ph.D., Professor, Pathology, University of Utah

Maximizing Reimbursement and How Coding Changes have Impacted a Complex Diagnostic Landscape
Instructors: Bonnie Ancone, Vice President, Molecular Diagnostics, XIFIN, Inc.
Kyle Fetter, Associate Vice President, Molecular Diagnostics, XIFIN, Inc.
Rina Wolf, Vice President, Commercialization Strategies, XIFIN, Inc.

DINNER

SUNDAY, FEBRUARY 9 | 5:30 – 8:30 PM

Starting an NGS Lab Part II: Practical and Business Aspects
Co-Organized with AMP Association for Molecular Pathology
Instructors: Victoria M. Pratt, Ph.D., FACMG, Director, Pharmacogenomics Laboratory, Medical & Molecular Genetics, Indiana University School of Medicine
Jill Hagenkord, M.D., CMO & Senior Vice President, InVitae
Roger Klein, M.D., J.D., Attending Pathologist, Molecular Pathology, Cleveland Clinic Foundation

NGS Assembly and Alignment
Instructor: Gabe Rudy, Vice President, Product Development, Golden Helix

PCR Part II: Digital PCR Applications and Advances
Instructor to be Announced, Inostics
Instructor to be Announced, LGC

Regulatory Compliance in Drug-Diagnostics Co-Development
Instructors: Maham Ansari, MS, RAC, Regulatory Affairs Consultant, US Strategic Regulatory Services, OptumInsight
Melina Cimler, Ph.D., Vice President, Quality & Regulatory, Illumina, Inc.

Building an Investigational Program to Establish the Clinical Value of a Test
Instructors: Catherine Schnabel, Ph.D., Vice President, Clinical Development & Medical Affairs, bioTheranostics, Inc.
Brock Schroeder, Ph.D., Director, Medical & Scientific Affairs, bioTheranostics, Inc.

MORNING

MONDAY, FEBRUARY 10 | 8:30 – 11:30 AM

Commercialization Boot Camp: Manual for Success in the Molecular Diagnostics Marketplace
Instructors: Harry Glorikian, Managing Director, Strategy, Precision for Medicine
Elaine Cheung, Business & Corporate Development, Illumina

Next-Generation Sequencing in Molecular Pathology: Challenges and Applications
Co-Organized with cap
Instructors: Wayne Grody, M.D., Ph.D., FCAP, FACMG, Professor, Departments of Pathology & Laboratory Medicine, Pediatrics, and Human Genetics, University of California, Los Angelese School of Medicine
Karl Voelkerding, M.D., Associate Professor, Pathology, University of Utah; Medical Director, Advanced Technology and Bioinformatics, ARUP Laboratories
Nazneen Aziz, Ph.D., Director, Molecular Medicine, Transformation Program Office, College of American Pathologists

Systems Biology: A Molecular Nutrition Perspective
Instructors: Jim Kaput, Ph.D., Head, Clinical Translation Unit, Nestle Institute of Health Sciences
Patrick J. Stover, Ph.D., Professor and Director, Division of Nutritional Sciences, Cornell University
Corrado Priami, Ph.D., President and CEO, The Microsoft Research - University of Trento Centre for Computational and Systems Biology (COSBI)

Neuro-Innovation
Instructors: Amit Etkin, M.D., Ph.D., Assistant Professor, Department of Psychiatry and Behavioral Sciences, Stanford University
Ruth O'Hara, Ph.D., Associate Professor (Research) of Psychiatry and Behavioral Sciences; Associate Director, Sierra-Pacific Mental Illness Education and Clinical Center, and (MIRECC) Director, National Fellowship Program in Advanced Psychiatry and Psychology Department, Stanford University School of Medicine
General Peter Chiarelli, United States Army (Ret.), Chief Executive Officer, One Mind for Research
Jeffrey S. Grethe, Ph.D., Co-Principal Investigator, Neuroscience Information Framework; Center for Research in Biological Systems School of Medicine, University of California, San Diego
Martin Kohn, MS, MD, FACEP, FACPE, Chief Medical Scientist, Care Delivery Systems, IBM Research

Isolation and Characterization of Cancer Stem Cells
Instructors to be Announced

The Next Frontier in Sample Shipping for Molecular Infectious Disease Testing: Eliminate Dry Ice
Instructor: Anita M. McClernon, Director of Laboratory Services, bioMONTR, a division of McClernon LLC

Genetically Engineered Mouse Models versus Patient-Derived Xenograft Models: Comparing Strengths and Limitations
Instructors: Serguei Kozlov, Ph.D., Principal Scientist, Center for Advanced Preclinical Research, SAIC-Frederick, Inc.
Additional Instructors to be Announced

Ethical, Legal, and Social Issues Related to Human Specimen Research
Instructors: Nicole Sieffert, Program Director, Biorepository Regulatory Support, MD Anderson Cancer Center
Marianna J. Bledsoe, Individual Consultant, Former Senior Program Manager, Department of Veterans Affairs, Office of Research and Development

Short Courses Maximize Your Productivity

Continued training and education are essential for staying competitive. Molecular Med Tri-Con Short Courses are designed to be instructive and interactive. These courses are a great introduction for those who are new to a particular discipline or as a refresher for those who want to brush up on their knowledge or expand their horizons. Attendance is limited to ensure an interactive environment. Group discussions are a key component in which course participants will have the opportunity to ask questions of the expert instructors and other participants. Course materials are included.

Short Courses are held prior to the main conference events so you won't miss a moment of TRI-CON 2014.

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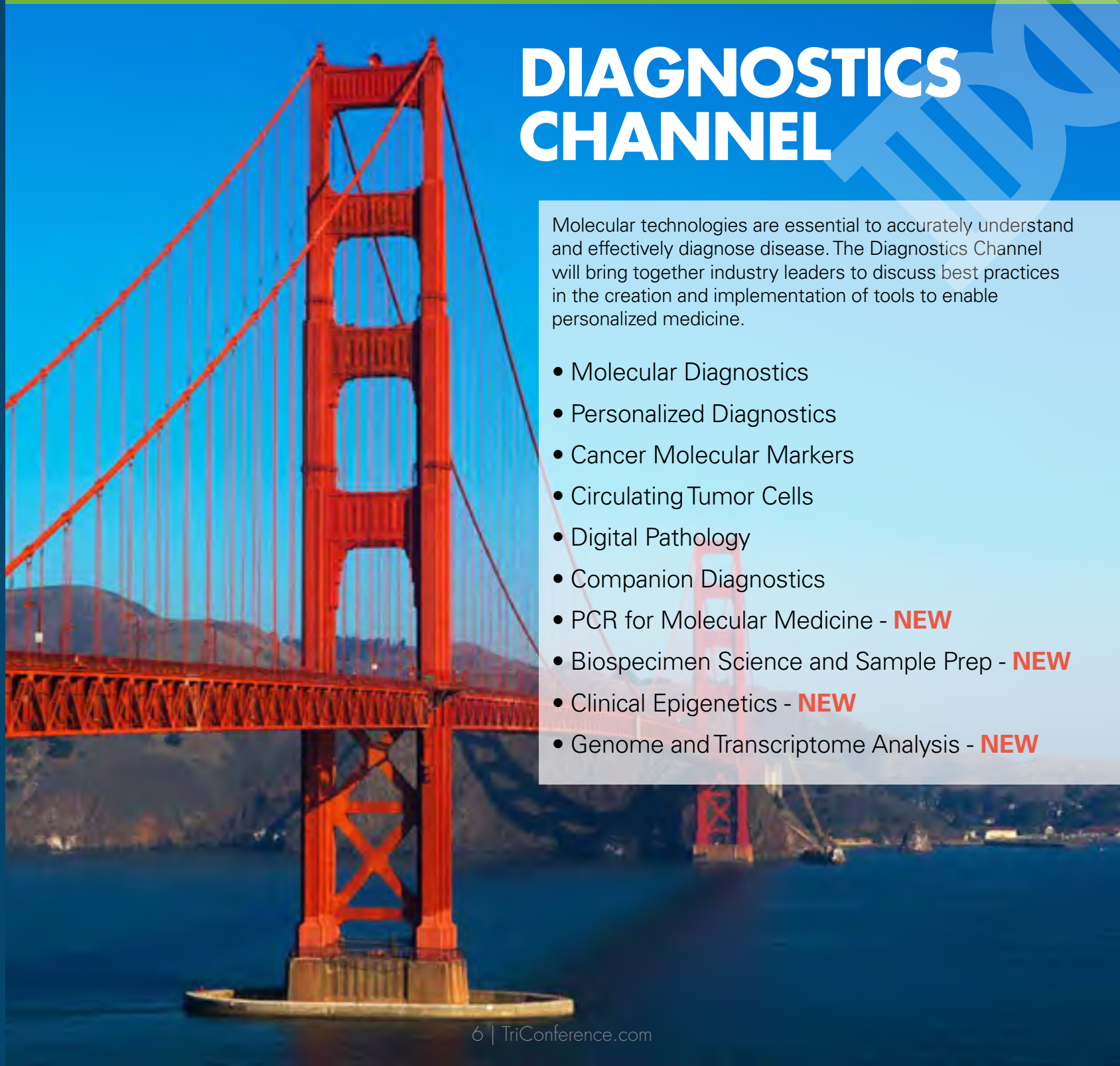

DIAGNOSTICS CHANNEL

Molecular technologies are essential to accurately understand and effectively diagnose disease. The Diagnostics Channel will bring together industry leaders to discuss best practices in the creation and implementation of tools to enable personalized medicine.

- Molecular Diagnostics
- Personalized Diagnostics
- Cancer Molecular Markers
- Circulating Tumor Cells
- Digital Pathology
- Companion Diagnostics
- PCR for Molecular Medicine - **NEW**
- Biospecimen Science and Sample Prep - **NEW**
- Clinical Epigenetics - **NEW**
- Genome and Transcriptome Analysis - **NEW**

The Industry Leader's Networking Event

Cambridge Healthtech Institute's Eleventh Annual

MOLECULAR DIAGNOSTICS

Executive Strategies for Success

MONDAY, FEBRUARY 10

10:30 am Conference Program Registration

11:50 Chairperson's Opening Remarks

Julien N. Bradley, MBA, Senior Director, Sales & Marketing, Quanterix Corporation

» 12:00 pm KEYNOTE PANEL DISCUSSION:

CPT Coding

Co-Organized with AMP Association for Molecular Pathology

Moderator: Jill Hagenkord, M.D., CMO & Senior Vice President, InVitae (2013 AMP Economic Affairs Committee and 2014 AMP Training and Education Committee)

Elaine Lyon, Ph.D., Medical Director, Molecular Genetics, ARUP (2014 AMP President and 2013 AMP Economic Affairs, Professional Relations, Strategic Opportunities Committees)

Chris L. Jagmin, M.D., Senior Medical Director, National Medical Policy and Operations, Aetna

- Overview of CPT codes
- Transitioning to the molecular pathology codes from the clinical laboratory's perspective
- Understanding rationale behind new CPT codes

1:00 Session Break

1:15 Luncheon Presentation I: Simoa HD-1: A Fully Automated, Multiplexed Immunoanalyzer with Single Molecule Sensitivity

Sponsored by Quanterix

David C. Duffy, Ph.D., CTO, Quanterix Corporation

Single Molecule Array (Simoa) technology allows multiple proteins to be detected at concentrations 1000-fold lower than currently possible. Simoa is based on the capture of single molecules on paramagnetic beads, and their detection in arrays of femtoliter wells. We will illustrate the power of this analytical sensitivity in diagnosing cancer, neurological diseases, and infectious diseases using the Simoa HD-1 Analyzer, a fully automated instrument designed for use in clinical research and diagnosis.

1:45 Luncheon Presentation II A Novel Exosome RNA Extraction Platform Enabling Biomarker Discovery and Diagnostic Development for Personalized Medicine

Sponsored by exosome diagnostics

Johan Skog, Ph.D., CSO, Exosome Diagnostics

Exosomes are released by cells as an active process of communication, and contain stable, intact nucleic acids, making them an ideal source for biomarker discovery and diagnostic development. ExoRNeasy was optimized to extract RNA, including mRNA, microRNAs, and other RNAs, from plasma and serum. The high quality RNA generated from the ExoRNeasy kit enables profiling of tumor associated mutations as well as RNA levels in biofluids of cancer patients.

2:15 Session Break

2:30 PANEL DISCUSSION: Opportunities and Challenges in Commercializing Esoteric Diagnostics

Co-Organized with PMC Personalized Medicine Coalition

Moderator: Edward Abrahams, Ph.D., President, Personalized Medicine Coalition

Organized by the Personalized Medicine Coalition, this panel will outline both the promise and the pitfalls in bringing new molecular diagnostics to market, including the scientific, regulatory, payment, and adoption issues that innovative diagnostics companies must negotiate. Featuring leaders in the field, it will provide insight into best practices and likely scenarios for future trends.

Peter Maag, Ph.D., CEO, XDx, Inc.

Steven Rosenberg, Ph.D., CSO, CardioDx, Inc.

Jay Wohlgemuth, M.D., Senior Vice President, Science & Innovation, Quest Diagnostics

Alan Wright, M.D., MPH, CMO, Roche Diagnostics

3:45 Next-Gen Sequencing for Prostate Cancer

Sponsored by ResearchDX Your Companion in Diagnostics

Philip D. Cotter, Ph.D., FACMG, FFSc(RCPA), Principal, ResearchDx

Diagnostic and prognostic assessment of prostate cancer has been dramatically improved by the use of new laboratory technologies. This presentation focuses on the use of gene panel and pathway analyses using Next-Gen sequencing approaches in prostate cancer.

4:00 Models for the Development of Multiplex Companion Diagnostics

Sponsored by ALMAC

Austin Tanney, Ph.D., Scientific Liaison Manager, Almac

4:15 A Bioinformatics Framework for Clinical Research Sequencing

Sponsored by life technologies™

Daniel Rhodes, Ph.D., Head, Medical Science Informatics, Life Technologies

The widespread adoption of precision medicine in oncology requires: a compendium of therapies targeting the genetic vulnerabilities of cancer; the diagnostic tools capable of generating a precise molecular diagnosis; and importantly, the information systems to connect a patient's molecular diagnosis to optimal treatment hypotheses. Here, we will present Life Technologies' work to accelerate (1) defining the landscape of actionable driver genetic events in cancer, (2) developing a next-generation sequencing screen on the Ion Torrent semiconductor-sequencing platform, (3) assembling treatment-related information relevant for clinical research, and (4) devising a turnkey bioinformatics solution to democratize clinical sequencing in the future.

4:30 Refreshment Break and Transition to Plenary Keynote

» 5:00 PLENARY KEYNOTE SESSION
(please see page 4 for details)

6:15 Grand Opening Reception in the Exhibit Hall with Poster Viewing

7:45 Close of Day

TUESDAY, FEBRUARY 11

7:00 am Registration and Morning Coffee

» 8:00 PLENARY KEYNOTE SESSION
(please see page 4 for details)

9:15 Refreshment Break in the Exhibit Hall with Poster Viewing

Sponsored by

10:15 A Few Crisp Comments by a Former FDA Leader on the Current Need for More FDA Reform Similar to What We Did Before

John A. Norris, J.D., MBA, Chairman, Norris Capital, Inc. and FDDH, Inc.; Senior US Advisor to Kanagawa, Japan, Governor Kuroiwa and GCC; Former 2nd-in-Command of the US FDA

This presentation discusses the efforts of the Global Collaborative Center (GCC) in helping to reform the U.S. FDA'S review methods and standards relating to diagnostics and other medical solutions like drugs and devices.

10:25 REGULATORY PANEL DISCUSSION: Late-Breaking News from the FDA that Impacts *in vitro* Diagnostic Tests

Moderator: Thomas F. Soriano, President & CEO, DOCRO, Inc.

MOLECULAR DIAGNOSTICS
CONTINUED ON NEXT PAGE

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MOLECULAR DIAGNOSTICS

continued

This session will feature guest speakers from the FDA and industry who will discuss late-breaking news related to companion diagnostics (CDx), next-generation sequencing, (NGS), laboratory-developed tests (LDTs), including guidance documents. Coordination of the pharmaceutical and diagnostic components for approval will be discussed - questions having to do with the process for approval of drugs that require a CDx, what to do with the various data, and how to anticipate requirements during the clinical stages of development will be discussed.

Guest Speaker via Webex: Alberto Gutierrez, Ph.D., Director, Office of in Vitro Diagnostics (OIR), FDA

Richard Naples, Senior Vice President, Worldwide Corporate Regulatory Affairs, Becton-Dickinson

Michael Page, Ph.D., Senior Director, Oncology, Global Regulatory Affairs, Eisai, Inc.

Daniel J. O'Shannessy, Ph.D., Senior Director, Translational Medicine & Diagnostics, Morphotek

12:00 pm Optimizing Evidence Development and Communication to Support Market Access for Molecular Diagnostics

Joseph V. Ferrara, President, Boston Healthcare

12:30 Session Break

12:40 Luncheon Presentation I: Personalized Patient Therapy with Pharmacogenetics

Sponsored by Luminex

Stuart A. Scott, Ph.D., Assistant Professor, Genetics and Genomic Sciences, Icahn School of Medicine at Mount Sinai

1:10 Luncheon Presentation II Washington Legal Update – Strategies for Navigating Key Reimbursement and FDA Issues for Diagnostics

Sponsored by Sidley Austin LLP

Torrey Cope, J.D., Partner, FDA Regulatory, Sidley Austin LLP

Barbara Cammarata, J.D., M.P.H., Counsel, Health Care Regulatory/ Reimbursement, Sidley Austin LLP

Dora L Hughes, M.D., M.P.H., Senior Policy Advisor, Government Strategies, Sidley Austin LLP

This session will feature an FDA lawyer, a health care reimbursement/CLIA/fraud and abuse lawyer, and a government strategies expert who will discuss some of the key legal and policy issues facing diagnostics companies in the current regulatory environment. The panel will explain practical strategies companies may employ to address legal challenges related to commercializing and marketing their products under FDA and health care requirements, as well as strategies to obtain appropriate reimbursement and to comply with government and private payor reimbursement requirements. It will also cover the industry's role in developing law and policy to address the unique challenges in the emerging areas of *in vitro* diagnostics, companion diagnostics, molecular pathology and next generation sequencing.

1:40 Refreshment Break in the Exhibit Hall with Poster Viewing

NOVEL TREATMENT STRATEGIES USING MOLECULAR DIAGNOSTICS: COMPANION DIAGNOSTIC OPPORTUNITIES

2:15 Chairperson's Remarks

William G. Loudon, M.D., Ph.D., Assistant Professor, Neurosurgery, University of California, Irvine; Section Chief, Neurosurgery, Children's Hospital of Orange County

2:20 Role of microRNAs in Signaling Pathways of Brain Tumor Malignancy

Sean Lawler, Ph.D., Assistant Professor, Dept. of Neurosurgery, Brigham and Women's Hospital, Harvard Medical School

2:50 BRAF Mutations

Leonard S. Sender, M.D., Medical Director, Hyundai Cancer Institute, CHOC Children's; Medical Director, Clinical Operations and Program Development, UC Irvine Medical Center Chao Family Comprehensive Cancer Center

BRAF mutations have been identified in a number of human cancers and diseases including melanoma, non-small cell lung cancer, papillary thyroid cancer, colorectal cancer, Langerhans cell histocytosis, myeloma, and hairy cell leukemia. This molecular target has been particularly intriguing to oncologists as there are FDA-approved targeted molecular agents available for use. This presentation will demonstrate how the lessons learned from the use of BRAF inhibitors may be applied to identify novel treatment strategies in other clinical settings.

3:20 Ongoing Phase III Clinical Trial at St. Joseph Hospital of DCVax-L: An Autologous Cellular Therapy for Treating Glioblastoma Multiforme

William G. Loudon, M.D., Ph.D., Assistant Professor, Neurosurgery, University of California, Irvine; Section Chief, Neurosurgery, Children's Hospital of Orange County

This talk will review the ongoing clinical trial of DCVax-L. The primary purpose of the study is to determine the efficacy of an investigational therapy called DCVax-L in patients with newly diagnosed GBM for whom surgery is indicated. The process, treatment and outcome will be discussed.

3:50 Developing Multiplexed Laboratory Developed Tests Using nCounter Elements™, a Novel Digital Barcoding Chemistry

Sponsored by nanoString Technologies

Joseph M. Beechem, Ph.D., Senior Vice President, Research & Development, NanoString Technologies

Translational genomics research has progressed rapidly to identifying clinically relevant gene expression, CNV and gene fusion signatures. nCounter Elements™ is a new, FFPE compatible, General Purpose Reagent (GPR) that enables these signatures to be rapidly implemented as Laboratory Developed Tests.

4:20 4:20 MRIGlobal Dx- National Solutions and Innovation, Worldwide Impact, Enabling Diagnostic Development for Patient Care

Sponsored by MRIGlobal

Meeta Patnaik, M.D., Medical Advisor, MRIGlobalDx

MRIGlobalDx enables medical device, platform, and technology companies to deliver products for patient care faster, more efficiently. With decades of assay development for agencies, including DOD and HHS, we facilitate commercial partners' diagnostic development and commercialization in today's healthcare environment.

4:35 Valentine's Day Celebration in the Exhibit Hall with Poster Viewing

5:20 Breakout Discussions in the Exhibit Hall *(see website for details)*

6:30 Close of Day

WEDNESDAY, FEBRUARY 12

7:00 am Breakfast Presentation *(Sponsorship Opportunity Available)* **or Morning Coffee**

» **8:00 PLENARY KEYNOTE SESSION** *(please see page 4 for details)*

9:45 Refreshment Break and Poster Competition Winner Announced in the Exhibit Hall

BUILDING THE CASE FOR REIMBURSEMENT- PRACTICAL CASE STUDIES

10:35 Chairperson's Remarks

Alan B. Carter, President, MDx Consulting

10:40 ConfirmMDx for Prostate Cancer: Navigating a Dynamic and Challenging Reimbursement Landscape

Jan Groen, Ph.D., CEO, MDx Health

Molecular diagnostics have lead to a significant improvement in diagnosing, treating and managing patient care. Despite significant advancements, labs and MDx innovators are faced with challenges encompassing coding, billing, and attempting to obtain coverage and fair reimbursement. The evidence bar is rising, with the burden on the MDx laboratory to prove clinical utility. In this case study we discuss the

MOLECULAR DIAGNOSTICS
CONTINUED ON NEXT PAGE

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MOLECULAR DIAGNOSTICS

continued

challenging reimbursement path for a new epigenetic test for prostate cancer.

11:10 Found in Translation: Taking the Cardiac Biomarker ST2 from Discovery to Clinical Adoption

James V. Snider, Ph.D., President, Critical Diagnostics

This presentation will touch on the discovery research and original identification ST2 as a potential cardiac biomarker. The emphasis though will be on the effort to develop a validated assay and the clinical evidence to justify routine clinical use of ST2 as a cardiac disease patient management tool, and will conclude with a discussion of the regulatory and reimbursement landscape.

11:40 Better Clinical Outcomes through Cardiovascular Genomics

Lon Castle, M.D., Medical Science Liaison, Medical Affairs, CardioDx

As the upcoming tidal wave of genomic tests floods the market, the molecular diagnostic industry will be faced with convincing payers that a new test provides enough value to warrant reimbursement. This presentation will focus on how a company can navigate the evidence development process in order to sail through a payer's technology assessment committee. The importance of testing the waters with an internal economic forecast modeling the impact of coverage will also be discussed.

12:10 pm Optimize for Variant Calling Confidence: Impact of Sample Type and Analytical Methods on NGS Data Quality

Sponsored by DNA genotek

Milos Popovic, Technical Product Manager, Seven Bridges Genomics

Seven Bridges Genomics, in collaboration with DNA Genotek, will present data and best practises for variant calling based on whole genome sequencing of DNA from matched pairs of saliva collected and stabilized with Oragene vs blood. When introducing personalized medicine offerings it is imperative to consider all parts of the patient value chain from sample collection to processing and validated results. Optimizing the patient experience, ensuring quality samples and future-proofing lab analysis through high quality specimen inputs is key to enabling successful, scalable health solutions.

12:25 Luncheon Presentation: Meeting the Clinical Utility Needs of Payers and Regulators for Molecular Diagnostics

Sponsored by PRECISION for MEDICINE

David Parker, Ph.D., Vice President, Market Access Strategy, Precision for Medicine

Judi Smith, Vice President, in vitro Diagnostics and Quality, Precision for Medicine

1:00 Refreshment Break in the Exhibit Hall and Last Chance for Poster Viewing

CHANGING CLINICAL DECISION SUPPORT BASED ON MOLECULAR DIAGNOSTICS

1:40 Chairperson's Remarks

Harry Glorikian, Managing Director, Strategy, Precision for Medicine

» 1:45 KEYNOTE PRESENTATION:

New Frontiers in Medical Diagnostics

Carlos Cordon-Cardo, M.D., Ph.D., Professor & Chair Pathology; Professor Genetics and Genomic Sciences; Professor Oncological Sciences, Mount Sinai Hospital

2:15 EXPERT PANEL DISCUSSION

With the dramatic increase in data being generated by molecular diagnostic devices (NGS in particular), very few clinical decisions are "binary" in nature today. Further, new technologies and clinical findings (mutations, pathways, drug resistance profiles) are being introduced regularly. To help compensate for the complexity and data overload, clinicians are hopeful that clinical decision support systems will meet the challenge. The panel will discuss their aspirations for these systems and whether existing solutions are solving their needs.

Panelists: Carlos Cordon-Cardo, M.D., Ph.D., Professor & Chair Pathology; Professor Genetics and Genomic Sciences; Professor Oncological Sciences, Mount Sinai Hospital

Kathryn A. Teng, M.D., FACP, Director, Center for Personalized Healthcare, Staff Physician, Internal Medicine, Cleveland Clinic; Assistant Professor, CWRU Lerner College of Medicine

Christopher D. Gocke, M.D., Associate Professor of Pathology and Oncology, Director, Heme Molecular Diagnostics, Johns Hopkins University School of Medicine

EU REGULATORY KEYNOTE

3:15 Chairperson's Remarks

Richard A. Montagna, Ph.D., Senior Vice President, Scientific Affairs, Rheonix, Inc.

3:20 Diagnostics Regulation and Policy in the EU

Hans-Georg Eichler, M.D., Senior Medical Officer, European Medicines Agency (EMA)

3:45 Refreshment Break

BIG DATA DRIVING PERSONALIZED MEDICINE

4:00 Chairperson's Remarks

Dalia Cohen, Ph.D., Founder & President, ALN Associates

4:05 Big Data's Big Role in Understanding Complex Diseases

Andreas Kogelnik, M.D., Ph.D., Founder and Director, Open Medicine Institute

The Open Medicine Institute (OMI) is effectively applying a collaborative, "big data" approach to understand and address complex diseases including: Autism, Lyme, Chronic Fatigue Syndrome, Parkinson's, and various cancers. This presentation will discuss the creation of a patient-centric infrastructure that handles and analyzes genomic sequencing information, bio-sampling data, physiology tests, basic research, patient experiences and physician evaluations to deliver needed information about a range of diseases.

4:35 Progress on Aggregating all the World's Genetic Tests into a Single Assay

Randy Scott, Ph.D., CEO and Co-Founder, InVitae

Technology is moving rapidly to enable massively parallel genetic testing. The ability to sequence DNA, however, is only the first step in building the infrastructure to analyze, store, manage, and interpret medical genetic information for patients. InVitae is focused on building the infrastructure to bring more comprehensive genetic testing into routine medical practice throughout the world.

5:05 It's Not Just About Big Data...Big Analytics for Identifying What Works and for Whom in Healthcare

Iya Khalil, Ph.D., Executive Vice President and Co-Founder, GNS Healthcare

We are living in the era of big data in healthcare, with unprecedented ability to collect data at multiple levels (genomic/'omic', phenotypic, health records, mobile health, etc.) and at scale. The key will be leveraging advanced analytics and appropriate feedback loops to identify what works on an individual patient level.

5:35 Close of Conference Program

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Cambridge Healthtech Institute's Fifth Annual

PERSONALIZED DIAGNOSTICS

Best Practices for Clinical Implementation

MONDAY, FEBRUARY 10

10:30 am Conference Program Registration

11:50 Chairperson's Opening Remarks
Julien N. Bradley, MBA, Senior Director, Sales & Marketing, Quanterix Corporation

» 12:00 pm KEYNOTE PANEL DISCUSSION:

CPT Coding

Co-Organized with AMP Association for Molecular Pathology

Moderator: Jill Hagenkord, M.D., CMO & Senior Vice President, InVitae (2013 AMP Economic Affairs Committee and 2014 AMP Training and Education Committee)
Elaine Lyon, Ph.D., Medical Director, Molecular Genetics, ARUP (2014 AMP President and 2013 AMP Economic Affairs, Professional Relations, Strategic Opportunities Committees)
Chris L. Jagmin, M.D., Senior Medical Director, National Medical Policy and Operations, Aetna

- Overview of CPT codes
- Transitioning to the molecular pathology codes from the clinical laboratory's perspective
- Understanding rationale behind new CPT codes

1:00 Session Break

1:15 Luncheon Presentation I: Applications of Biomarkers in Oncology Clinical Development

Sponsored by GENOPTIX BIOPHARMA SERVICES

Jelveh Lameh, Ph.D., Executive Director, BioPharma Services, Genoptix, Inc., A Novartis Company

The desire for personalized cancer treatments is increasing with the advent of new technologies and the changing regulatory landscape. This presentation will highlight applications of biomarker assays that have been developed and utilized in clinical trials for exploratory use, pharmacodynamic assessments, target enrichment, and patient preselection and stratification. Case studies utilizing genetic and protein tissue biomarkers in clinical trials will be discussed.

1:45 Luncheon Presentation II *(Sponsorship Opportunity Available)*

2:15 Session Break

PAST, PRESENT, AND FUTURE OF PRENATAL GENOMIC TESTING

2:30 Chairperson's Remarks
Daniel H. Farkas, Ph.D., HCLD, FACB, Laboratory Director, Sequenom Center for Molecular Medicine

2:35 Noninvasive Prenatal Testing by Maternal Plasma DNA Sequencing: From Aneuploidy Testing to Fetal Whole Genome and Methylome Sequencing
Dennis Lo, M.D., Ph.D., Director, Li Ka Shing Institute of Health Sciences, Chinese University of Hong Kong

Over the last 5 years, there is much interest in the use of massively parallel sequencing of plasma DNA for noninvasive prenatal testing. Fetal chromosomal aneuploidies, genome-wide molecular karyotyping and even fetal whole genome sequencing were accomplished through the sequencing of maternal plasma DNA. We have shown that this approach can be used for noninvasive determination of the fetal methylome. This latter development has opened exciting opportunities for prenatal testing and research.

3:05 Noninvasive Fetal RHD Genotyping
Daniel H. Farkas, Ph.D., HCLD, FACB, Laboratory Director, Sequenom Center for Molecular Medicine

Antepartum anti-D immunoprophylaxis is standard of care in pregnancy management but is administered unnecessarily to the approximately 40% of mothers who subsequently deliver Rh-negative babies. Clinical laboratory investigation using circulating, cell-free fetal DNA as an analyte provides the potential for more rational management of Rh-negative pregnant women.

3:35 Noninvasive Prenatal Testing 2014: The Basics and Beyond
Christopher Robinson, M.D., MSCR, Associate Professor, Maternal Fetal Medicine, Obstetrics and Gynecology, University of Virginia

This presentation will present the current, state-of-the-art in applied screening and diagnostics involving cffDNA. A focus on the understanding of integration of bench science and bioinformatics will provide the attendee with an expanded understanding of the utilization of cffDNA as a substrate for clinical information and decision-making.

4:05 Clinician-Friendly Tools and Efficient Database Architecture to Accelerate Genetic Diagnoses of Challenging Patients

Sponsored by NEXTCODE HEALTH

Jeff Gulcher, M.D., Ph.D., President, CSO, NextCODE Health

NextCODE offers informatics systems originally developed at deCODE Geneticsalong with a curated knowledge base for end-to-end analysis of whole exome and whole genome data for patients, families, and large cohorts. Clinical Sequence Analyzer (CSA) is a clinician-friendly web-based interface for sequence-based diagnostics. NextCODE's GOR database infrastructure allows for more efficient storage, query, and display of large scale sequence and coverage data. Our *de novo* mutation detector improves accuracy of *de novo* mutation calls.

4:35 Refreshment Break and Transition to Plenary Keynote

» 5:00 PLENARY KEYNOTE SESSION
(please see page 4 for details)

6:15 Grand Opening Reception in the Exhibit Hall with Poster Viewing

7:45 Close of Day

TUESDAY, FEBRUARY 11

7:00 am Registration and Morning Coffee

» 8:00 PLENARY KEYNOTE SESSION
(please see page 4 for details)

9:15 Refreshment Break in the Exhibit Hall with Poster Viewing

Sponsored by BIOTECHNOLOGY EXECUTIVE SEARCH

10:25 REGULATORY PANEL DISCUSSION: Late-Breaking News from the FDA that Impacts *in vitro* Diagnostic Tests
Moderator: Thomas F. Soriano, President & CEO, DOCRO, Inc.

This session will feature guest speakers from the FDA and industry who will discuss late-breaking news related to companion diagnostics (CDx), next-generation sequencing, (NGS), laboratory-developed tests (LDTs), including guidance documents. Coordination of the pharmaceutical and diagnostic components for approval will be discussed - questions having to do with the process for approval of drugs that require a CDx, what to do with the various data, and how to anticipate

PERSONALIZED DIAGNOSTICS
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PERSONALIZED DIAGNOSTICS

continued

requirements during the clinical stages of development will be discussed.

Guest Speaker via Webex: Alberto Gutierrez, Ph.D., Director, Office of in Vitro Diagnostics (OIR), FDA

Richard Naples, Senior Vice President, Worldwide Corporate Regulatory Affairs, Becton-Dickinson

Michael Page, Ph.D., Senior Director, Oncology, Global Regulatory Affairs, Eisai, Inc.

Daniel J. O'Shannessy, Ph.D., Senior Director, Translational Medicine & Diagnostics, Morphotek

12:00 pm Optimizing Evidence Development and Communication to Support Market Access for Molecular Diagnostics

Joseph V. Ferrara, President, Boston Healthcare

Learn how the landscape for molecular diagnostics is evolving in terms of market access pathways and clinical evidence requirements. Attendees will gain an understanding of the challenges of aligning evidence development programs to support clinical positioning, market adoption, and payer reimbursement and funding. Best practices for collaborations between clinical development and commercialization strategy teams will be reviewed and highlighted, as well as key elements of successful transitions from clinical development to commercialization and payer engagement.

12:30 Session Break

12:40 Luncheon Presentation I: Personalized Patient Therapy with Pharmacogenetics

Sponsored by Luminex

Stuart A. Scott, Ph.D., Assistant Professor, Genetics and Genomic Sciences, Icahn School of Medicine at Mount Sinai

1:10 Luncheon Presentation II Washington Legal Update – Strategies for Navigating Key Reimbursement and FDA Issues for Diagnostics

Sponsored by SIDLEY AUSTIN LLP SIDLEY

Torrey Cope, J.D., Partner, FDA Regulatory, Sidley Austin LLP

Barbara Cammarata, J.D., M.P.H., Counsel, Health Care Regulatory/ Reimbursement, Sidley Austin LLP

Dora L Hughes, M.D., M.P.H., Senior Policy Advisor, Government Strategies, Sidley Austin LLP

This session will feature an FDA lawyer, a health care reimbursement/CLIA/fraud and abuse lawyer, and a government strategies expert who will discuss some of the key legal and policy issues facing diagnostics companies in the current regulatory environment. The panel will explain practical strategies companies may employ to address legal challenges related to commercializing and marketing their products under FDA and health care requirements, as well as strategies to obtain appropriate reimbursement and to comply with government and private payor reimbursement requirements. It will also cover the industry's role in developing law and policy to address the unique challenges in the emerging areas of *in vitro* diagnostics, companion diagnostics, molecular pathology and next generation sequencing.

1:40 Refreshment Break in the Exhibit Hall with Poster Viewing

2:15 STRATEGIC PANEL DISCUSSION: Recent Companion Diagnostic Drug Approvals

Moderator: Hakan Sakul, Ph.D., Executive Director and Head, Diagnostics, Worldwide R&D, Clinical Research and Precision Medicine; La Jolla Site Head, Development Operations, Pfizer, Inc.

Overview on diagnostic commercialization, reading market forces and what it takes to make a success.

Ron Mazumder, Ph.D., MBA, Global Head, Research & Product Development, Janssen/J&J

Peter Collins, Ph.D., Vice President & Head, Diagnostics, GlaxoSmithKline

Jeremy Bridge-Cook, Ph.D., Senior Vice President, Research & Development, Luminex

Eric Lai, Ph.D., Senior Vice President & Head, Pharmacogenomics, Takeda Pharmaceuticals

Gregory Zdechlik, COO, Eli Lilly & Co.

3:50 Sponsored Presentations *(Opportunities Available)*

4:20 Valentine's Day Celebration in the Exhibit Hall with Poster Viewing

5:20 Breakout Discussions in the Exhibit Hall *(see website for details)*

6:30 Close of Day

WEDNESDAY, FEBRUARY 12

7:00 am Breakfast Presentation *(Sponsorship Opportunity Available)* **or Morning Coffee**

» **8:00 PLENARY KEYNOTE SESSION**
(please see page 4 for details)

9:45 Refreshment Break and Poster Competition Winner Announced in the Exhibit Hall

DIFFERENT APPROACHES TO DIAGNOSING USING NGS

10:35 Chairperson's Remarks

Karl Voelkerding, M.D., Associate Professor, Pathology, University of Utah; Medical Director, Advanced Technology & Bioinformatics, ARUP Laboratories

10:40 Use of Exome Sequencing for Genetic Diagnosis: Clinical Experience and Case Examples

Wayne W. Grody, M.D., Ph.D., Professor, Medical Genetics and Molecular Pathology, Pathology & Lab Medicine, Pediatrics, and Human Genetics; Director, Molecular Diagnostic Laboratories and Clinical Genomics Center, UCLA School of Medicine

The advent of massively parallel or "next-generation" DNA sequencing has finally brought into reach the long-anticipated "Thousand Dollar Genome", or the ability to sequence an individual's entire genome at reasonable cost. This presentation will review such aspects as clinical utility, challenges in test interpretation and genetic counseling, return of incidental findings and reimbursement, all within the context of our own experience performing clinical whole-exome sequencing at an academic medical center.

11:10 The Art of Interpreting and Reporting Results from a Multi-Gene, NGS-Based Panel for Solid Tumor Mutation Testing

Allie Grossmann, M.D., Ph.D., Staff Pathologist, Surgical Pathology and Oncology, ARUP Laboratories

With the advent of clinical testing of tumors with NGS technology, the breadth of sequence coverage has expanded beyond the scope of well-characterized mutations. This presents a challenge to both interpreting variants and to meeting reasonable clinical turn-around times for reporting. These challenges will be discussed within the context of clinical case examples with an emphasis on "lessons learned" in the adoption of NGS for solid tumor mutation testing.

11:40 Providing More Comprehensive Genetic Diagnostics by Next-Generation Sequencing-Based Multi-Gene Panels

Karl Voelkerding, M.D., Associate Professor, Pathology, University of Utah; Medical Director, Advanced Technology & Bioinformatics, ARUP Laboratories

For many genetic disorders, heterogeneity is the rule and NGS has provided a new avenue by which to overcome the limitations of Sanger sequencing-based diagnostic approaches. This presentation will highlight how NGS is transforming the diagnostic evaluation of genetic disorders through more comprehensive multi-gene panel-based methods. Technical options and bioinformatics considerations will be discussed along with how Sanger sequencing and NGS are being used in an integrative fashion.

12:10 pm Pertinence Metric Enables Hypothesis-Independent Genome-Phenome Analysis in Seconds

Sponsored by SimulConsult

Michael M. Segal, M.D., Ph.D., Chief Scientist, SimulConsult

Genome-phenome analysis uses a genomic variant table and compares patient's findings to those of known diseases ("phenome"). Accuracy was 100% with trios with family-aware calling, and close to that with only probands. The gene pertinence metric calculated in the analysis was 99.9%

PERSONALIZED DIAGNOSTICS
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for the causal genes, and the analysis took seconds and was hypothesis-independent.

12:25 Luncheon Presentation: Sequencer-Ready Target Enrichment for Clinical Next Generation Sequencing: A Massively Parallel Singleplex PCR Approach

Sponsored by WAFERGEN BIOSYSTEMS

Xun Xu, Ph.D., Deputy Director, BGI Research, WaferGen BioSystems

Advances in NGS technology have resulted in dramatic improvements in sequencing throughput and turnaround time, yet a critical bottle neck in NGS workflows exists at the library preparation stage. We present a technology that dramatically reduces process time by generating sequencer-ready amplicon libraries in a single step. This coupled with the ability to simultaneously process multiple samples enables a high fidelity, scalable and cost effective high throughput solution for NGS targeted resequencing.

1:00 Refreshment Break in the Exhibit Hall and Last Chance for Poster Viewing

INSIGHTS INTO BIOLOGY OF CANCER FROM NGS

1:40 Chairperson's Remarks

German Pihan, M.D., Director, Hematopathology Lab, Pathology, Beth Israel Deaconess Medical Center & Harvard Medical School

1:45 The Spectrum of Mutations and Mutated Cancer Genes Across Many Tumor Types

Michael Lawrence, Ph.D., Computational Biologist, Cancer Genome Computational Analysis, Broad Institute of MIT and Harvard University

Comprehensive knowledge of the genes underlying human cancers is a critical foundation for cancer diagnostics, therapeutics, clinical trial design and selection of rational combination therapies. While some cancer genes are mutated at very high frequency (>20% of patients), the vast majority are found at intermediate frequencies (2-20%) and sometimes even lower frequencies.

2:15 The Evolving Genome of Glioblastoma

Siyuan Zheng, Ph.D., Postdoctoral Fellow, Bioinformatics & Computational Biology, MD Anderson Cancer Center, University of Texas

Glioblastoma (GBM) rates amongst the most deadly of adult tumors. We have performed extensive genomic profiling to characterize the landscape of somatic alterations of GBM. Analysis of matched pre- and post-treatment GBM showed that the GBM genome evolves under the selective pressures from cytotoxic therapy and cytoreductive surgery. We provide new insights into the heterogeneity of this disease, and the factors contributing to it.

2:45 Mutational Heterogeneity and Evolution in Diffuse Large B-Cell Lymphoma

Ryan D. Morin, Ph.D., Assistant Professor, Bioinformatics, Molecular Biology and Biochemistry, Simon Fraser University

Diffuse large B-cell lymphoma is the most common aggressive non-Hodgkin lymphoma. Whole genome and transcriptome sequencing in this tumor type has uncovered a plethora of common mutation targets and a long tail of infrequently mutated genes with potential relevance. We have also computationally dissected individual DLBCL tumors to identify multiple sub clones and evidence for ongoing acquisition of driver mutations in this disease.

3:15 Sponsored Presentations *(Opportunities Available)*

3:45 Refreshment Break

EVALUATING CANCER MOLECULAR MARKERS

4:00 Chairperson's Remarks

Daniel W. Chan, Ph.D., DABCC, FACB, Professor, Pathology, Oncology, Radiology and Urology; Director, Center for Biomarker Discovery and Translation & Clinical Chemistry, Johns Hopkins University

» 4:05 KEYNOTE PRESENTATION:

Key to Success in Translating Cancer Biomarkers to the Clinical Laboratory

Daniel W. Chan, Ph.D., DABCC, FACB, Professor, Pathology, Oncology, Radiology and Urology; Director, Center for Biomarker Discovery and Translation & Clinical Chemistry, Johns Hopkins University

Specific examples will be shown to demonstrate the opportunities and challenges for the development of clinical proteomic diagnostics. The successful translation of these biomarkers into clinical practice will require close collaboration between researcher, industry, regulatory agency and clinician.

4:35 Advanced Machine Learning Techniques Reveal Molecular Correlates of Tumor Histopathology in GBM and Low Grade Glioma

Bahram Parvin, Ph.D., Principal Scientist, Life Sciences, Lawrence Berkeley National Laboratory

The case study includes Glioblastoma Multiform from The Cancer Genome Atlas (TCGA), where four subtypes were identified from computed morphometric indices, and a subset of these subtypes are shown to be predictive of the outcome. In each case, molecular correlates are able to provide hypotheses for therapeutic targeting.

5:05 Clinical Evaluation of Exosomes as Oncology Biomarkers

Shidong Jia, Ph.D., Scientist, Oncology Biomarker Development, Genentech, Inc.

Dr. Jia's lab has developed working procedures to evaluate exosomal RNA signature as novel biomarkers for cancer prognosis, prediction and patient stratification. In particular, their work has refreshed current practice and demonstrated a new approach for studying RNA signature in patient blood samples.

5:35 Close of Conference Program

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CANCER MOLECULAR MARKERS

Guiding Cancer Management

MONDAY, FEBRUARY 10

10:30 am Conference Program Registration

» OPENING KEYNOTE SESSION: CRITERIA FOR VALIDATION

11:50 Chairperson's Opening Remarks

12:00 pm Clinical Validation

Howard I. Scher, M.D., D. Wayne Calloway Chair in Urologic Oncology, Sidney Kimmel Center for Prostate and Urologic Cancers; Chief, Genitourinary Oncology Service, Memorial Sloan-Kettering Cancer Center

Recent insights into the molecular basis of prostate cancer led to the successful development of rationally designed therapies targeting the androgen receptor (AR) and AR signaling axis. Not all patients respond, suggesting the presence of predictive biomarkers of sensitivity. Circulating tumor cells (CTCs) provide a representation of the genetic makeup of an individual patient's tumor. Most studies analyze cells in bulk, limiting the ability to identify molecular changes present in smaller populations that may be associated with *de novo* or acquired resistance. Single-cell genomic analyses avoid this limitation, and an opportunity to better guide management.

12:30 Validation in Clinic and Trial: Why So Hard?

George W. Sledge Jr., M.D., Chief, Oncology, Medicine, Stanford University School of Medicine

1:00 Session Break

1:15 Luncheon Presentation I

Sponsored by Janssen

Mark Connelly, Ph.D., Scientific Director, Cellular Research, Janssen R&D

1:45 Luncheon Presentation II

Sponsored by BIOSCALE

Blood-Based Strategies to Monitor Disease and Response to Targeted Therapies

Cloud Paweletz, Ph.D., Head, Translation Research Laboratory, Belfer Institute for Applied Cancer Science, Dana Faber Cancer Institute

2:15 Session Break

CIRCULATING BIOMARKERS - WHICH IS BEST FOR LIQUID BIOPSY?

2:30 Chairperson's Remarks

Klaus Pantel, M.D., Professor & Founding Director, Institute of Tumor Biology, University Medical Center Hamburg-Eppendorf, University of Hamburg

2:35 Prognostic Indicators in Peripheral Blood of Dukes' Stage B Colorectal Cancer Cases

Koshi Mimori, M.D., Ph.D., Director & Professor, Surgery, Kyushu University Beppu Hospital, Japan

We identified EMT inducible gene Plastin3 (PLS3), an actin bundling protein. PLS3 in the peripheral blood is a suitable new marker for CTCs that has strong and independent prognostic significance in CRC, especially in Dukes' Stage B patients, who have the strongest need for improved risk assessment.

3:05 Detection and Characterization of Viable Circulating Tumor Cells

Catherine Alix-Panabières, Ph.D., Maître de Conférence, Praticien Hospitalier, Associate Professor, Director, Laboratory of Rare Human Circulating Cells, Institute of Research in Biotherapy, Saint-Eloi Hospital, University Medical Centre of Montpellier

The enumeration and characterization of circulating tumor cells (CTCs) in the peripheral blood and disseminated tumor cells (DTCs) in bone marrow may provide important prognostic information and might help to monitor efficacy of therapy. Since current assays cannot distinguish between apoptotic and viable DTCs/CTCs, it is now possible to apply a novel ELISPOT assay (designated 'EPISPOT') that detects proteins secreted/released/shed from single epithelial cancer cells.

3:35 CTC vs. ctDNA – Which is More Informative?

Klaus Pantel, M.D., Professor & Founding Director, Institute of Tumor Biology, University Medical Center Hamburg-Eppendorf, University of Hamburg

The analysis of therapeutic targets and drug resistance-conferring gene mutations on CTCs and ctDNA opens new perspectives in cancer therapy. The current challenges of CTCs and ctDNA as biomarkers in clinical oncology will be discussed. Both CTCs and ctDNA are complementary technologies that can be used in parallel in future trials assessing new drugs or drug combinations.

4:05 Comparing Genomic Mutations from Solid Tumors and CTCs: A Case Study

Sponsored by silicon biosystems

Nicolo Manaresi, Ph.D., Technology Officer, Silicon Biosystems S.p.A.

The paper will present preliminary results from a study in which mutational status of individual tumor cells recovered from tissue biopsies are compared to the mutational profiles of CTCs recovered from the same patient.

4:20 High Precision Microfilters Accurately Identify True Circulating Tumor Cells and Other Cancer Markers

Sponsored by CREATV MicroTech

Cha-Mei Tang, Sc.D., President & CEO, Creatv MicroTech, Inc.

Precision microfilters provide rapid and efficient capture of CTCs and other biomarkers. The filtration system provides gentle and easy workflow. Cells are well preserved enabling high-definition fluorescent imaging to accurately identify CTCs by morphology. Single cells can be extracted for analysis. CTCs can be cultured directly in the filtration system.

4:35 Refreshment Break and Transition to Plenary Keynote

» 5:00 PLENARY KEYNOTE SESSION
(please see page 4 for details)

6:15 Grand Opening Reception in the Exhibit Hall with Poster Viewing

7:45 Close of Day

TUESDAY, FEBRUARY 11

7:00 am Registration and Morning Coffee

» 8:00 PLENARY KEYNOTE SESSION
(please see page 4 for details)

9:15 Refreshment Break in the Exhibit Hall with Poster Viewing

Sponsored by HIGHI PARTNERS EXECUTIVE SEARCH

CANCER MOLECULAR MARKERS
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CANCER MOLECULAR MARKERS

continued

NEW MARKERS

10:25 Chairperson's Remarks
Steven A. Soper, Ph.D., Biomedical Engineering, Chemistry; William H. Pryor Emeritus Professor, Director, Center for BioModular Multi-Scale Systems, University of North Carolina, Chapel Hill

10:30 Single-Cell Analyses Identifying EMT-Related Biomarkers for Metastatic Prostate Cancer
Chun-Liang Chen, Ph.D., Assistant Professor/Research, Molecular Medicine, Institute of Biotechnology, Cancer Therapy and Research Center, University of Texas Health Science Center at San Antonio
Single-cell approaches are used to study EMT-related markers of CTCs for metastatic prostate cancer detection and prediction. Markers are identified using microfluidics and atomic force microscope (AFM) for molecular profiling and nanomechanical and nanochemical phenotypes of CTCs, respectively.

11:00 Integrated System for the Efficient Analysis of CTC Sub-populations with Divergent Phenotypes
Steven A. Soper, Ph.D., Biomedical Engineering, Chemistry; William H. Pryor Emeritus Professor, Director, Center for BioModular Multi-Scale Systems, University of North Carolina, Chapel Hill
Rare CTC sub-populations can be isolated directly from whole blood using the appropriate markers and processed for molecular signatures to identify key drivers of, for example, epithelial-to-mesenchymal transitions that may be associated with spawning metastatic disease. New microfluidic systems and assays for processing CTC sub-populations and their molecular signatures will be discussed.

11:30 Characterizing Breast Cancer CTCs for Brain Metastasis Competence
Dario Marchetti, Ph.D., Professor, Pathology & Immunology and Molecular & Cellular Biology; Director, CTC Core Facility, Baylor College of Medicine
We report novel strategies investigating breast cancer CTCs isolated from peripheral blood mononuclear cells of patients with or without clinically diagnosed brain metastasis. We demonstrated the metastatic competency of CTCs and necessity of a specific CTC profile to promote brain metastasis. These results and strategies can have high impact towards developing new and effective therapies combating breast cancer metastasis in general, brain metastasis in particular.

12:00 pm Characterization of Circulating Large Cells Isolated by ScreenCell-Filtration in Renal Tumor Patients
Sponsored by ScreenCell
Karoline Lackner, M.D., Professor, Pathology, Institute of Pathology, Medical University of Graz
Circulating non-hematologic cells (CNHCs) - putative CTCs/CTMs - isolated from blood by ScreenCell filtration in 40 patients with renal tumors were characterized. We found that CNHC-clusters with malignant or uncertain malignant cytomorphological features were CD45neg/CD31pos. Array-CGH revealed a balanced genome in 83% of cytomorphologically malignant/uncertain malignant CNHC-clusters whereas 17% showed genomic DNA imbalances not found in the primary tumors. Thus the majority of CNHC-clusters may be of endothelial origin.

12:30 Enjoy Lunch on Your Own

1:40 Refreshment Break in the Exhibit Hall with Poster Viewing

CELL-FREE DNA

2:15 Chairperson's Remarks
Stefanie Jeffrey, M.D., John & Marva Warnock Professor, Surgery, Chief of Surgical Oncology Research, Stanford University School of Medicine

2:20 The Clinical Application of Circulating Tumor DNA
Luis A. Diaz, M.D., Associate Professor, Oncology, Johns Hopkins Sidney Kimmel Comprehensive Cancer Center
We developed a dynamic biomarker based on this premise utilizing highly sensitive digital PCR-based and next-generation sequencing-based assays. Ongoing efforts are expanding the role of ctDNA measurements in a variety of clinical scenarios and for the genotyping of patients enrolled in clinical trials. This technology is also being incorporated into the human clinical trials as a companion diagnostic measuring key predictive mutations in the blood.

2:50 Detection of Circulating Tumor DNA by Deep Sequencing
Maximilian Diehn, M.D., Ph.D., Assistant Professor, Radiation Oncology, Stanford Cancer Institute, Institute for Stem Cell Biology & Regenerative Medicine, Stanford University
This presentation will discuss detection of circulating tumor DNA using deep sequencing and its potential clinical applications.

3:20 Clinical Utility of Cell-Free DNA/miRNA: Present and Future
David Hoon, MSc, Ph.D., Director, Molecular Oncology, John Wayne Cancer Institute
Cell-free DNA (cfDNA) has evolved into a provocative new approach of assessing genomic and epigenomic alterations in cancer patients' blood. In recent years, cfDNA has been shown to be a potential blood biomarker for monitoring tumor progression.

3:50 Genomic Testing in Oncology: Toward Truly Personalized Cancer Management
Sponsored by illumina
Daniel S. Grosu, M.D., MBA, CMO, Diagnostics, Illumina Inc.
Rapid advances in next-generation sequencing technology are now enabling routine interrogation of tumor genetics on an unprecedented scale. From deep targeted panels to whole genome sequencing, innovative research tools are providing critical new insights into tumor biology, paving the way for earlier diagnosis and more personalized treatment. Emerging methods based on circulating tumor DNA are particularly exciting as they hold the promise of noninvasive tumor detection and characterization, with potential applications across the continuum of cancer care.

4:20 Valentine's Day Celebration in the Exhibit Hall with Poster Viewing

5:20 Breakout Discussions in the Exhibit Hall *(see website for details)*

6:30 Close of Day

WEDNESDAY, FEBRUARY 12

7:00 am Breakfast Presentation *(Sponsorship Opportunity Available)* **or Morning Coffee**

8:00 PLENARY KEYNOTE SESSION
(please see page 4 for details)

9:45 Refreshment Break and Poster Competition Winner Announced in the Exhibit Hall

EPIGENETIC BIOMARKERS AND DIAGNOSTICS

10:35 Chairperson's Remarks
Michelle M. Hanna, Ph.D., CEO & Scientific Director, RiboMed Biotechnologies, Inc.

10:40 Identification of Genome-Wide Methylated CpG Island Profiles of Coding, Non-Coding and Repeated Regions as Molecular Markers for Human Melanomas
Ranjan J. Perera, Ph.D., Associate Professor & Scientific Director, Genomics and Bioinformatics, Sanford-Burnham Medical Research Institute

CANCER MOLECULAR MARKERS
CONTINUED ON NEXT PAGE

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CANCER MOLECULAR MARKERS

continued

We have identified genome-wide methylated CpG island distributions by subjecting melanoma genomic DNA to methyl binding domain 2 (MBD2) pull-down and NGS. CpG islands in the upstream regulatory regions of many coding and non-coding RNA genes exhibit extensive hypermethylation, whereas several repeated elements, such as LINE 2, and several LTR elements, are hypomethylated in advanced stage melanoma cell lines. Focused assays of melanoma patient tissue samples for CpG island methylation near the noncoding RNA genes demonstrated high specificity.

11:10 miRNA Biomarkers for Colorectal Neoplasia

Ajay Goel, Ph.D., Director, Epigenetics and Cancer Prevention, Baylor Research Institute

MicroRNAs (or miRNAs) are small transcripts of 20-24 nucleotides that have emerged as important regulators of gene expression in cancer cells. Overexpression of specific miRNAs has been linked to the stepwise disease progression during the normal-adenoma-cancer sequence in the colorectal cancer (CRC). Given their cancer-specific pattern of expression, remarkable stability and presence in blood and other body fluids, miRNAs are considered to be highly promising cancer biomarkers.

11:40 PANEL DISCUSSION: Developing and Commercializing Epigenetic Diagnostics

Moderator:

Perry Dimas, VP, Business Development, Premier Source Diagnostics

Panelists:

Michelle M. Hanna, Ph.D., CEO & Scientific Director, RiboMed Biotechnologies, Inc.

Noel Doheny, CEO, Epigenomics, Inc.

Babak Alizadeh, Ph.D., Co-founder & COO, PrognosDx Health, Inc.

Additional Panelists to be Announced

Epigenetic marks, such as DNA methylation and histone modifications, comprise part of the epigenetic machinery leading to abnormal gene expression and chromatin instability in disease. Epigenetic changes, particularly in human cancers, are now being considered as novel biological markers for diagnostic and therapeutic utility. This panel will discuss current challenges in developing and commercializing epigenetic diagnostics by addressing three specific areas of concern:

- DNA Methylation as Viable Biomarkers
- Validation of Assay and Technologies
- CPT Coding and Reimbursement Challenges

12:10 pm Session Break

12:40 Luncheon Presentations *(Sponsorship Opportunities Available)* **or Lunch on Your Own**

1:40 Refreshment Break in the Exhibit Hall with Poster Viewing

PRE-ANALYTICAL CONSIDERATIONS IN CANCER GENOMIC ANALYSIS

1:40 Chairperson's Remarks

Jane Emerson, M.D., Ph.D., Professor of Clinical Pathology, University of Southern California

1:45 Biospecimen Quality Appraisal at a Cancer Tissue Bank

Teri A. Longacre, M.D., Professor of Pathology; Director, Tissue, Procurement Facility, Stanford Cancer Center

Quality control of biospecimen banking in a high volume academic cancer center requires a dedicated anatomic pathologist with specific expertise in tumor pathology. Methods to ensure high quality biospecimen banking, storage, and distribution based on best practices and evidence-based standards are presented.

2:15 Quantification of HER2 Heterogeneity in Patient Samples

Elena Geretti, Ph.D., Senior Scientists, Merrimack Pharmaceuticals

The currently FDA-approved methods for HER2 quantification are not able to give a quantitative measure of HER2 heterogeneity of expression. We have developed a novel immunofluorescence assay to quantify HER2 on FFPE samples. Coupled with automated image analysis, our assay is able to quantify HER2 expression at the single cell level, and may constitute a means to understand the predictive and/or prognostic role of the heterogeneity of HER2 expression for patient response to HER2-targeted therapies.

2:45 Sample Type Bias in the Analysis of Cancer Genomes: How Admixed Normal Cells, Intratumoral Heterogeneity, and *ex vivo* Growth Impact Cancer Genomic Analyses

David Solomon, M.D., Ph.D., Anatomic Pathology, PGY-2, University of California, San Francisco

Techniques have emerged which now allow us to interrogate the entire genome of human cancers and to define the genetic alterations that drive tumorigenesis. When performing these genomic analyses, it is important to understand the impact that admixed normal cells, intratumoral heterogeneity, and *ex vivo* growth can have on the results. This talk will discuss these issues and highlight one study comparing genomic analyses performed on glioblastoma tumor samples of differing types (primary tumors, primary xenografts, primary cultures, and established cell lines).

3:15 Sponsored Presentations *(Opportunities Available)*

3:45 Refreshment Break

EVALUATING CANCER MOLECULAR MARKERS

4:00 Chairperson's Remarks

Daniel W. Chan, Ph.D., DABCC, FACB, Professor, Pathology, Oncology, Radiology and Urology; Director, Center for Biomarker Discovery and Translation & Clinical Chemistry, Johns Hopkins University

» 4:05 KEYNOTE PRESENTATION:

Key to Success in Translating Cancer Biomarkers to the Clinical Laboratory

Daniel W. Chan, Ph.D., DABCC, FACB, Professor, Pathology, Oncology, Radiology and Urology; Director, Center for Biomarker Discovery and Translation & Clinical Chemistry, Johns Hopkins University

Specific examples will be shown to demonstrate the opportunities and challenges for the development of clinical proteomic diagnostics. The successful translation of these biomarkers into clinical practice will require close collaboration between researcher, industry, regulatory agency and clinician.

4:35 Advanced Machine Learning Techniques Reveal Molecular Correlates of Tumor Histopathology in GBM and Low Grade Glioma

Bahram Parvin, Ph.D., Principal Scientist, Life Sciences, Lawrence Berkeley National Laboratory

The case study includes Glioblastoma Multiform from The Cancer Genome Atlas (TCGA), where four subtypes were identified from computed morphometric indices, and a subset of these subtypes are shown to be predictive of the outcome. In each case, molecular correlates are able to provide hypotheses for therapeutic targeting.

5:05 Clinical Evaluation of Exosomes as Oncology Biomarkers

Shidong Jia, Ph.D., Scientist, Oncology Biomarker Development, Genentech, Inc.

Dr. Jia's lab has developed working procedures to evaluate exosomal RNA signature as novel biomarkers for cancer prognosis, prediction and patient stratification. In particular, their work has refreshed current practice and demonstrated a new approach for studying RNA signature in patient blood samples.

5:35 Close of Conference Program

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Cambridge Healthtech Institute's Fourth Annual

CIRCULATING TUMOR CELLS

Spotlight on Clinical Validation

MONDAY, FEBRUARY 10

10:30 am Conference Program Registration

» OPENING KEYNOTE SESSION: CRITERIA FOR VALIDATION

11:50 Chairperson's Opening Remarks

12:00 pm Clinical Validation

Howard I. Scher, M.D., D. Wayne Calloway Chair in Urologic Oncology, Sidney Kimmel Center for Prostate and Urologic Cancers; Chief, Genitourinary Oncology Service, Memorial Sloan-Kettering Cancer Center

Recent insights into the molecular basis of prostate cancer led to the successful development of rationally designed therapies targeting the androgen receptor (AR) and AR signaling axis. Not all patients respond, suggesting the presence of predictive biomarkers of sensitivity. Circulating tumor cells (CTCs) provide a representation of the genetic makeup of an individual patient's tumor. Most studies analyze cells in bulk, limiting the ability to identify molecular changes present in smaller populations that may be associated with *de novo* or acquired resistance. Single-cell genomic analyses avoid this limitation, and an opportunity to better guide management.

12:30 Validation in Clinic and Trial: Why So Hard?

George W. Sledge Jr., M.D., Chief, Oncology, Medicine, Stanford University School of Medicine

1:00 Session Break

1:15 Luncheon Presentation I

Sponsored by Janssen

Mark Connelly, Ph.D., Scientific Director, Cellular Research, Janssen R&D

1:45 Luncheon Presentation II

Sponsored by BIOSCALE

Blood-Based Strategies to Monitor Disease and Response to Targeted Therapies

Cloud Paweletz, Ph.D., Head, Translation Research Laboratory, Belfer Institute for Applied Cancer Science, Dana Faber Cancer Institute

2:15 Session Break

CIRCULATING BIOMARKERS - WHICH IS BEST FOR LIQUID BIOPSY?

2:30 Chairperson's Remarks

Klaus Pantel, M.D., Professor & Founding Director, Institute of Tumor Biology, University Medical Center Hamburg-Eppendorf, University of Hamburg

2:35 Prognostic Indicators in Peripheral Blood of Dukes' Stage B Colorectal Cancer Cases

Koshi Mimori, M.D., Ph.D., Director & Professor, Surgery, Kyushu University Beppu Hospital, Japan

We identified EMT inducible gene Plastin3 (PLS3), an actin bundling protein. PLS3 in the peripheral blood is a suitable new marker for CTCs that has strong and independent prognostic significance in CRC, especially in Dukes' Stage B patients, who have the strongest need for improved risk assessment.

3:05 Detection and Characterization of Viable Circulating Tumor Cells

Catherine Alix-Panabières, Ph.D., Maître de Conférence, Praticien Hospitalier, Associate Professor, Director, Laboratory of Rare Human Circulating Cells, Institute of Research in Biotherapy, Saint-Eloi Hospital, University Medical Centre of Montpellier

The enumeration and characterization of circulating tumor cells (CTCs) in the peripheral blood and disseminated tumor cells (DTCs) in bone marrow may provide important prognostic information and might help to monitor efficacy of therapy. Since current assays cannot distinguish between apoptotic and viable DTCs/CTCs, it is now possible to apply a novel ELISPOT assay (designated 'EPISPOT') that detects proteins secreted/released/shed from single epithelial cancer cells.

3:35 CTC vs. ctDNA – Which is More Informative?

Klaus Pantel, M.D., Professor & Founding Director, Institute of Tumor Biology, University Medical Center Hamburg-Eppendorf, University of Hamburg

The analysis of therapeutic targets and drug resistance-conferring gene mutations on CTCs and ctDNA opens new perspectives in cancer therapy. The current challenges of CTCs and ctDNA as biomarkers in clinical oncology will be discussed. Both CTCs and ctDNA are complementary technologies that can be used in parallel in future trials assessing new drugs or drug combinations.

4:05 Comparing Genomic Mutations from Solid Tumors and CTCs: A Case Study

Sponsored by silicon biosystems

Nicolo Manaresi, Ph.D., Technology Officer, Silicon Biosystems S.p.A.

The paper will present preliminary results from a study in which mutational status of individual tumor cells recovered from tissue biopsies are compared to the mutational profiles of CTCs recovered from the same patient.

4:20 High Precision Microfilters Accurately Identify True Circulating Tumor Cells and Other Cancer Markers

Sponsored by

Cha-Mei Tang, Sc.D., President & CEO, Creatv MicroTech, Inc.

Precision microfilters provide rapid and efficient capture of CTCs and other biomarkers. The filtration system provides gentle and easy workflow. Cells are well preserved enabling high-definition fluorescent imaging to accurately identify CTCs by morphology. Single cells can be extracted for analysis. CTCs can be cultured directly in the filtration system.

4:35 Refreshment Break and Transition to Plenary Keynote

» 5:00 PLENARY KEYNOTE SESSION
(please see page 4 for details)

6:15 Grand Opening Reception in the Exhibit Hall with Poster Viewing

7:45 Close of Day

TUESDAY, FEBRUARY 11

7:00 am Registration and Morning Coffee

» 8:00 PLENARY KEYNOTE SESSION
(please see page 4 for details)

9:15 Refreshment Break in the Exhibit Hall with Poster Viewing

Sponsored by

NEW MARKERS

10:25 Chairperson's Remarks

Steven A. Soper, Ph.D., Biomedical Engineering, Chemistry; William H. Pryor Emeritus Professor, Director, Center for BioModular Multi-Scale Systems, University of North Carolina, Chapel Hill

CIRCULATING TUMOR CELLS
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continued

10:30 Single-Cell Analyses Identifying EMT-Related Biomarkers for Metastatic Prostate Cancer

Chun-Liang Chen, Ph.D., Assistant Professor/Research, Molecular Medicine, Institute of Biotechnology, Cancer Therapy and Research Center, University of Texas Health Science Center at San Antonio

Single-cell approaches are used to study EMT-related markers of CTCs for metastatic prostate cancer detection and prediction. Markers are identified using microfluidics and atomic force microscope (AFM) for molecular profiling and nanomechanical and nanochemical phenotypes of CTCs, respectively.

11:00 Integrated System for the Efficient Analysis of CTC Sub-populations with Divergent Phenotypes

Steven A. Soper, Ph.D., Biomedical Engineering, Chemistry; William H. Pryor Emeritus Professor, Director, Center for BioModular Multi-Scale Systems, University of North Carolina, Chapel Hill

Rare CTC sub-populations can be isolated directly from whole blood using the appropriate markers and processed for molecular signatures to identify key drivers of, for example, epithelial-to-mesenchymal transitions that may be associated with spawning metastatic disease. New microfluidic systems and assays for processing CTC sub-populations and their molecular signatures will be discussed.

11:30 Characterizing Breast Cancer CTCs for Brain Metastasis Competence

Dario Marchetti, Ph.D., Professor, Pathology & Immunology and Molecular & Cellular Biology; Director, CTC Core Facility, Baylor College of Medicine

We report novel strategies investigating breast cancer CTCs isolated from peripheral blood mononuclear cells of patients with or without clinically diagnosed brain metastasis. We demonstrated the metastatic competency of CTCs and necessity of a specific CTC profile to promote brain metastasis. These results and strategies can have high impact towards developing new and effective therapies combating breast cancer metastasis in general, brain metastasis in particular.

12:00 pm Characterization of Circulating Large Cells Isolated by ScreenCell-Filtration in Renal Tumor Patients

Sponsored by ScreenCell

Karoline Lackner, M.D., Professor, Pathology, Institute of Pathology, Medical University of Graz

Circulating non-hematologic cells (CNHCs) - putative CTCs/CTMs - isolated from blood by ScreenCell filtration in 40 patients with renal tumors were characterized. We found that CNHC-clusters with malignant or uncertain malignant cytomorphological features were CD45neg/CD31pos. Array-CGH revealed a balanced genome in 83% of cytomorphologically malignant/uncertain malignant CNHC-clusters whereas 17% showed genomic DNA imbalances not found in the primary tumors. Thus the majority of CNHC-clusters may be of endothelial origin.

12:30 Enjoy Lunch on Your Own

1:40 Refreshment Break in the Exhibit Hall with Poster Viewing

CELL-FREE DNA

2:15 Chairperson's Remarks

Stefanie Jeffrey, M.D., John & Marva Warnock Professor, Surgery, Chief of Surgical Oncology Research, Stanford University School of Medicine

2:20 The Clinical Application of Circulating Tumor DNA

Luis A. Diaz, M.D., Associate Professor, Oncology, Johns Hopkins Sidney Kimmel Comprehensive Cancer Center

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Maximilian Diehn, M.D., Ph.D., Assistant Professor, Radiation Oncology, Stanford Cancer Institute, Institute for Stem Cell Biology & Regenerative Medicine, Stanford University

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3:50 Genomic Testing in Oncology: Toward Truly Personalized Cancer Management

Sponsored by illumina

Daniel S. Grosu, M.D., MBA, CMO, Diagnostics, Illumina Inc.

Rapid advances in next-generation sequencing technology are now enabling routine interrogation of tumor genetics on an unprecedented scale. From deep targeted panels to whole genome sequencing, innovative research tools are providing critical new insights into tumor biology, paving the way for earlier diagnosis and more personalized treatment. Emerging methods based on circulating tumor DNA are particularly exciting as they hold the promise of noninvasive tumor detection and characterization, with potential applications across the continuum of cancer care.

4:20 Valentine's Day Celebration in the Exhibit Hall with Poster Viewing

5:20 Breakout Discussions in the Exhibit Hall *(see website for details)*

6:30 Close of Day

WEDNESDAY, FEBRUARY 12

7:00 am Breakfast Presentation *(Sponsorship Opportunity Available)* **or Morning Coffee**

8:00 PLENARY KEYNOTE SESSION *(please see page 4 for details)*

9:45 Refreshment Break and Poster Competition Winner Announced in the Exhibit Hall

EMERGING TECHNOLOGIES FOR CTC DETECTION AND ANALYSIS

10:35 Chairperson's Remarks

Avraham Rasooly, Ph.D., Program Director, NCI NIH

10:40 Isolation and Analysis of Individual Circulating Tumor Cells with eDAR

Daniel T. Chiu, Ph.D., A. Bruce Montgomery Professor of Chemistry & Bioengineering, University of Washington, Seattle

This presentation will describe a method we developed for isolating circulating tumor cells from peripheral blood, with emphasis on a set of probes that we have developed to enhance our ability to detect and isolate these rare cells. We will also describe the performance of this system, the downstream single-cell analysis we have performed, and its clinical utility.

11:10 High-Throughput CTC Capture, Sorting and Analysis Using a Velocity Valley Chip

Shana O. Kelley, Ph.D., Distinguished Professor, Chemistry, Biochemistry, Pharmaceutical Sciences, Biomedical Engineering, University of Toronto

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CIRCULATING TUMOR CELLS

continued

A new device for CTC isolation and analysis will be described that allows separation of different CTC populations and integrated genetic analysis. The characterization of this device with clinical samples and a comparison with the gold standard for CTC analysis will be presented.

11:40 Aptamers and DNA Nanospheres for Isolation of Cancer Cells Using Microfluidic Devices

Z. Hugh Fan, Ph.D., Professor, Interdisciplinary Microsystems Group, Mechanical & Aerospace Engineering, Biomedical Engineering, Chemistry, University of Florida

We will present our results on incorporating DNA aptamers with microfluidic devices for the isolation of cancer cells from whole blood. The performance of aptamers will be compared with DNA nanospheres, each of which is comprised of a gold nanoparticle conjugated with a number of aptamers and is capable of multivalent binding with a cancer cell. Isolation and enumeration of circulating tumor cells (CTC) from pancreatic cancer patients will also be discussed.

12:10 pm Session Break

12:20 Luncheon Presentation *(Sponsorship Opportunity Available)* **or Lunch on Your Own**

1:00 Refreshment Break in the Exhibit Hall and Last Chance for Poster Viewing

1:40 Chairperson's Remarks

Avraham Rasooly, Ph.D., Program Director, NCI NIH

1:45 Flexible Micro Spring Array Device (FMSA) for High-Throughput Enrichment of Viable Circulating Tumor Cells

Siyang Zheng, Ph.D., Assistant Professor, Bioengineering, Pennsylvania State University

The enrichment of viable CTCs for *ex vivo* analysis could further improve cancer diagnosis and guide treatment selection. The new flexible micro spring array (FMSA) device enriches viable CTCs independent of antigen expression. The FMSA device provides a versatile and reliable platform capable of viable enrichment and analysis of CTCs from clinically relevant volumes of whole blood.

2:15 Microfluidics for Characterization of Circulating Tumor Cells

Jiang F. Zhong, Ph.D., Assistant Professor, Pathology, University of Southern California

Microfluidic devices for isolation and molecular characterization of single cells will be discussed.

2:45 Microencapsulated Sensors for Circulating Tumor Cell Detection

Weian Zhao, Ph.D., Professor, Pharmaceutical Sciences, Sue and Bill Gross Stem Cell Research Center, Chao Family Comprehensive Cancer Center, University of California, Irvine

This is a new platform technology that surpasses some of the challenges faced by current CTC detection systems including sensitivity, specificity, and throughput. It will be of broad interest to academia, industry and patient care.

3:15 Sponsored Presentations *(Opportunities Available)*

3:45 Refreshment Break

NEW DEVICES

4:00 Chairperson's Remarks

Steven A. Soper, Ph.D., Biomedical Engineering, Chemistry; William H. Pryor Emeritus Professor, Director, Center for BioModular Multi-Scale Systems, University of North Carolina, Chapel Hill

4:05 MicroHall Sensor for Direct CTC Detection and Profiling in Blood

Hakho Lee, Ph.D., Assistant Professor, Center for Systems Biology, Massachusetts General Hospital, Harvard Medical School

This presentation will describe a novel, chip-based magnetic cytometer that can detect and molecularly profile CTCs directly from patient samples.

4:35 Measuring Physical Properties of Circulating Tumor Cells

Scott R. Manalis, Ph.D., Professor, Biological Engineering, Massachusetts Institute of Technology

My lab is developing microfluidic approaches for measuring multiple physical properties of single cells with high precision and high throughput. I will describe a new method we developed for characterizing the deformability and surface friction of cancer cells and will present recent progress on determining if this method can identify circulating tumor cells in cancer patient samples.

5:05 CTCs and Their Applications to Clinical Laboratories

Ali Asgar Bhagat, Ph.D., Technical Director, R&D, Clearbridge BioMedics Pte Ltd

Koh Furuta, M.D., Ph.D., Head, Clinical Laboratories, National Cancer Center Hospital

We utilized a prototype of CTC isolation device in collaboration with National Singapore University. This particular device can isolate CTCs on the microfluidic platform. We have successfully isolated CTCs derived from advanced colon cancer patients. A part of the isolated CTCs is utilized for biobanking and NGS.

5:35 Close of Conference Program

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Cambridge Healthtech Institute's Second Annual

DIGITAL PATHOLOGY

Transforming Medicine in a Digital World

MONDAY, FEBRUARY 10

10:30 am Conference Program Registration

» FEATURED PRESENTATIONS

11:50 Chairperson's Opening Remarks
Ralf Huss, M.D., Chief Medical Officer, Definiens AG

12:05 pm American Telemedicine Association (ATA) Updated Guideline on Telepathology
Liron Pantanowitz, M.D., Associate Professor, Pathology, University of Pittsburgh Medical Center
The practice of telepathology has evolved significantly. New technology has been developed, clinical and non-clinical applications have expanded, innovative business opportunities have arisen, and regulatory issues have emerged. The ATA accordingly convened a working group to develop updated guidelines to address these trends. This talk will review these new guidelines addressing issues regarding technology, clinical practice, applications, clinical and facility responsibilities, validation, training, reporting, quality assurance, maintenance, and security, as well as medicolegal, regulatory and ethical considerations.

12:35 Pushing the Resolution Limit with a Simple LED Array
Guoan Zheng, Ph.D., Assistant Professor, Biomedical Engineering & Electrical Engineering, University of Connecticut
Conventional slide scanner acquires multiple high-resolution images and stitches them in the spatial domain. We discuss an imaging modality, termed Fourier ptychographic microscopy (FPM), which acquires multiple low-resolution images and stitches them in the Fourier domain. We report a FPM prototype that uses a LED array and a 2X objective lens to achieve the resolution of a 20X objective lens.

1:05 Session Break

1:15 Luncheon Presentation I Pathology - From an Analog Art to a Digital Science
Sponsored by DEFINIENS Understanding Images
Ralf Huss, M.D., Chief Medical Officer, Definiens AG
Anatomical pathology has been evolving from a merely descriptive (analog) subject of post-mortem autopsies to a single cell-based analysis of molecular events that might even predict the response to targeted therapies. Now, tissue-based diagnostics reaches the next level of clinical utility with the quantitation of almost endless (big) data points and context-based digital information, mining all available sets of information from individual samples, including the clinical outcome, and translating it into "Tissue Phenomics."

1:45 Luncheon Presentation II *(Sponsorship Opportunity Available)*

2:15 Session Break

IN VIVO MICROSCOPY

2:30 Chairperson's Remarks
Eric F. Glassy, M.D., Medical Director, Pathology, Inc.

2:35 IVM Diagnosis in Surgical Pathology: Present and Future
Maria M. Shevchuk, M.D., Anatomic Pathology & Clinical Pathology, Weill Cornell Medical College
In vivo microscopy technologies are used as *in vivo* intraprocedural diagnostic tools, and for *ex vivo* applications in the surgical pathology. In the near future, these technologies will become an integral part of the practice of pathology. This lecture highlights the current uses, and addresses several "value added" applications for the future. The participants will be better able to introduce these technologies into their practice of surgical pathology.

3:05 *In vivo* Microscopy: Technologies, Applications and Roles for Pathologists
Guillermo J. Tearney, M.D., Ph.D., Mike & Sue Hazard Family MGH Research Scholar, Professor, Pathology, Harvard Medical School; Wellman Center for Photomedicine, Massachusetts General Hospital

3:35 *In vivo* Imaging of Cell and Tissue Dynamics: Towards Biologically Relevant Diagnostic Models in Surgical Pathology
Kamran Badizadegan, M.D., FCAP, Chair, Pathology & Laboratory Medicine, Nemours Children's Hospital; Professor of Pathology, UCF College of Medicine
Surgical pathology, considered the gold standard in anatomical diagnosis, is by and large a subjective pattern recognition exercise in which excised, chemically fixed, thinly sectioned and stained tissues are examined by a bright field light microscope. Although this practice has served medicine well for decades, it ignores the dynamical cell and tissue processes that define health and disease. This presentation will build on the general concept of *in vivo* microscopy (IVM) to make a case for cell and tissue dynamics as potential diagnostic criteria.

4:05 An Objective Image Quality Model Based on Subjective Pathology Perception Test
Sponsored by PHILIPS
Dirk Vossen, Ph.D., Philips Digital Pathology

4:20 Sponsored Presentation *(Opportunity Available)*

4:35 Refreshment Break and Transition to Plenary Keynote

» 5:00 PLENARY KEYNOTE SESSION
(please see page 4 for details)

6:15 Grand Opening Reception in the Exhibit Hall with Poster Viewing

7:45 Close of Day

TUESDAY, FEBRUARY 11

7:00 am Registration and Morning Coffee

» 8:00 PLENARY KEYNOTE SESSION
(please see page 4 for details)

9:15 Refreshment Break in the Exhibit Hall with Poster Viewing
Sponsored by CHI PARTNERS EXECUTIVE SEARCH

MULTIPLEXING

10:25 Chairperson's Remarks
Kenneth J. Bloom, M.D., CMO, Clarient, Inc.

10:30 Overview of Multiplexing and Novel Microscopy Platforms: So Many Biomarkers, So Little Tissue…
Richard Levenson, M.D., Professor and Vice Chair, Strategic Technologies, Pathology & Laboratory Medicine, University of California, Davis Medical Center
Pathology is evolving to accommodate an explosion of molecular markers being used for diagnostic, prognostic, and therapy-guidance purposes. At the same time, minimally invasive procedures are generating ever smaller tissue samples to be analyzed. Fortunately, new multiplexing reagents, improved microscope optics and sensors, and advanced software tools can help.

DIGITAL PATHOLOGY
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DIGITAL PATHOLOGY

continued

11:00 Predicting Response to Targeted Therapy in Solid Tumors Using Multivariate Indices based on Protein Biomarker Expression

Vladimir Knezevic, M.D., Senior Vice President, Research & Development, 20/20 GeneSystems, Inc.

Companion diagnostics are expected to guide future treatment decisions by identifying the patients most likely to benefit from a particular (often highly priced) cancer therapeutic. Also, by ruling out therapies that are not likely to be effective, this approach could save unnecessary costs and improve standard of care. 20/20 GeneSystems, Inc. is utilizing unique layered immunohistochemistry technology to develop assays for analysis of multiple protein biomarkers in solid tumors.

11:30 Multiplexing in a CLIA Environment

Kenneth J. Bloom, M.D., CMO, Clarient, Inc.

The size of biopsy samples is getting smaller and smaller the number of actionable therapeutic targets is increasing. There are many practical aspects to consider when implementing multiplexing procedures and there are even more considerations when implementing a complex test in a CLIA environment. I will review our experience with implementing a 9-plex immunostain in our clinical lab and discuss issues likely to be encountered when expanding the IHC panels to include DNA and RNA probes.

12:00 Sponsored Presentations *(Opportunities Available)*

12:30 Session Break

12:40 Luncheon Presentations *(Sponsorship Opportunities Available)* **or Lunch on Your Own**

1:40 Refreshment Break in the Exhibit Hall with Poster Viewing

QUANTITATIVE BIOMARKERS

2:15 Chairperson's Remarks

David L. Rimm, M.D., Ph.D., Professor, Pathology, Yale University

2:20 The Quest for a Universal Fixative: Measuring Fixative-induced Morphologic and Antigenic Variation

Alexander "Sandy" Borowsky, M.D., Center for Comparative Medicine, University of California, Davis

Quantitative Image Analysis (QIA) for morphometric parameters and immunohistochemistry of breast cancer antigens was used to evaluate the technical reproducibility, biological variability, and intratumoral heterogeneity in animal tumor models and compared to human clinical samples in several ways. This talk will review the findings on the technical reproducibility and biological heterogeneity as well as the results of fixatives in both morphometric and immunohistochemical parameters.

2:50 New Methods for Multi-Parameter Quantitative *in situ* Biomarker Assessment: Nucleic Acids vs. Proteins

David L. Rimm, M.D., Ph.D., Professor, Pathology, Yale University

While *in situ* protein and DNA measurements (quantitative or semi-quantitatively) have been done for years, *in situ* hybridization (ISH) for RNA has historically been challenging. On the other hand, the types and functions for RNA, now including microRNA and long non-coding (linc) RNA have expanded dramatically over the last few years. Here we describe methods for quantitative nucleic acid measurement and discuss comparisons between RNA measurements by RT-PCR vs. quantitative ISH.

3:20 Mass Tags and IHC—A New Frontier for 100 Parameters and Above

Garry P. Nolan, Ph.D., Rachford and Carlota A. Harris Professor, Microbiology & Immunology, Stanford School of Medicine; Director, NHLBI Proteomics Center for Systems Immunology; Baxter Laboratory for Stem Cell Biology, Center for Clinical Science Research

Multiparameter single cell analysis (MPSCA) for histological and non-adherent cells has been a critical mainstay of clinical diagnostic procedures for decades. Recent innovations using mass spectrometry tags for single cell analysis in conjunction with inductively coupled mass spectrometry and secondary ion mass spectrometry is radically altering analysis frontiers. I will discuss such innovations and informatics approaches to data analysis and representation of results from clinical samples in such high dimensionality arenas.

4:20 Valentine's Day Celebration in the Exhibit Hall with Poster Viewing

5:20 Breakout Discussions in the Exhibit Hall *(see website for details)*

6:30 Close of Day

WEDNESDAY, FEBRUARY 12

7:00 am Breakfast Presentation *(Sponsorship Opportunity Available)* **or Morning Coffee**

» **8:00 PLENARY KEYNOTE SESSION**
(please see page 4 for details)

9:45 Refreshment Break and Poster Competition Winner Announced in the Exhibit Hall

IMPLEMENTATION & PRACTICE

10:35 Chairperson's Remarks

Liron Pantanowitz, M.D., Associate Professor, Pathology, University of Pittsburgh Medical Center; Pathology, UPMC Shadyside

10:40 Management of Clinical Digital Image Files

John H. Sinard, M.D., Ph.D., Professor, Pathology; Director, Pathology & Informatics; Associate Director, Anatomic Pathology, Yale University

Pathology departments accumulate digital image files of all different kinds. Proper management of these files is important to assure their availability. Appropriate consideration needs to be given to the design of the image storage process since the initial solution chosen is likely to persist for years. This talk will discuss many of the issues that need to be considered in designing a clinical digital image management solution.

11:10 Implementing WSI in a Large Academic Hospital

Thomas W. Bauer, M.D., Ph.D., Medical Director, ePathology, Cleveland Clinic

Successful implementation of WSI for research, education, and patient care in a large academic department requires acceptance among more than just early adopters. This talk will review our strategy to develop, implement and measure a dynamic "digital education library," and the outcome of the secondary diagnosis validation study.

11:40 Image Analysis Algorithms: What Can They Do for Pathology?

Metin N. Gurcan, Ph.D., Associate Professor, Biomedical Informatics, Ohio State University

The qualitative analysis of histopathological images is a time-consuming process and is subject to inter- and intra-reader variations. This often negatively affects the diagnosis, prognosis and treatment of patients. In this talk, we will discuss how computer-assisted interpretation

DIGITAL PATHOLOGY
CONTINUED ON NEXT PAGE

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DIGITAL PATHOLOGY

continued

of histopathological slides can assist pathologists in their decision-making.

12:10 pm Session Break

12:20 Luncheon Presentation *(Sponsorship Opportunity Available)* **or Lunch on Your Own**

1:00 Refreshment Break in the Exhibit Hall and Last Chance for Poster Viewing

IMAGE APPLICATIONS

1:40 Chairperson's Remarks
Liron Pantanowitz, M.D., Associate Professor, Pathology, University of Pittsburgh Medical Center; Pathology, UPMC Shadyside

1:45 Use of Whole Slide Imaging for Primary Diagnosis in Canada
Andrew J. Evans, M.D., Ph.D., FRCPC, Staff Pathologist & Associate Professor, University Health Network, Laboratory Medicine Program

There is growing acceptance for using WSI for diagnostic purposes, including consultations and frozen sections. Interest in Canada and other countries is also building around the use of this technology for primary diagnosis, particularly in multi-site institutions with consolidated pathology departments and subspecialty sign out. This presentation will review logistical, regulatory and medicolegal factors that facilitate the use of WSI for primary diagnosis in these situations in Canada.

2:15 Digital Imaging of Peripheral Blood Smears: Challenges and Opportunities
Christopher Naugler, M.D., BScH, MSc, CCFP, FCFP, FRCPC, Assistant Professor, Pathology and Laboratory Medicine, University of Calgary, Division Head, General Pathology, Calgary Laboratory Services

Digital imaging of peripheral blood smears, combined with image analysis software is becoming commonplace in clinical laboratories. The single cell nature of blood image analysis is analogous to cytologic image analysis but still has room for improvement in accuracy. New algorithms (especially when applied in parallel) may improve image analysis accuracy.

2:45 Implementation of WSI for Consensus Review in Clinical Trials
Stephen M. Hewitt, M.D., Ph.D., Clinical Investigator, Laboratory of Pathology, Center for Cancer Research, National Cancer Institute

The implementation of WSI in a clinical trial setting offers a critical view of the challenges in implementation of the technology into the pathology routine. This presentation will discuss the advantages of the adoption of WSI in a clinical trial, as well as the considerations and technical task of implementation for a successful integration of digital pathology in clinical trials.

3:15 How Molecular Pathology Blends with Laboratory Medicine
George D. Lundberg, M.D., Chief Medical Officer & Editor in Chief, CollabRx

3:45 Refreshment Break

ADVANCED IMAGING TECHNIQUES

4:00 Chairperson's Remarks
Anil Parwani, M.D., Ph.D., Director, Pathology Informatics, Pathology, University of Pittsburgh Medical Center

4:05 Why Investing in Digital Imaging is a Good Bet
Anil Parwani, M.D., Ph.D., Director, Pathology Informatics, Pathology, University of Pittsburgh Medical Center

As pathology practices consider if they should invest in digital pathology or not, they will encounter difficult decisions surrounding the cost of the technology vs. the benefits. This lecture will provide an overview of some clinical applications of digital pathology and the types of uses of this technology that would be potentially beneficial to the practices in spite of the high costs of the technology.

4:35 Alterations in Nanoscale Nuclear Architecture for Cancer Diagnosis and Prognosis
Yang Liu, Ph.D., Assistant Professor, Medicine, Bioengineering, Hillman Cancer Center, University of Pittsburgh

Alteration in nuclear architecture is one of the hallmarks in cancer diagnosis and prognosis, but the diffraction-limited resolution of conventional light microscopy limits their accuracy in some scenarios of cancer diagnosis and prognosis. Our group developed a novel optical microscopy system to interrogate 3D nanoscale alterations in nuclear architecture using clinically prepared routine tissues and cells. We will present their use to improve cancer diagnosis and prognosis in multiple cancers.

5:05 Whole Slide Image Analysis in Fine Needle Aspiration Biopsy and Cytopathology
Brian Collins, Ph.D., Associate Professor, Pathology and Immunology, Washington University, St. Louis

Whole slide images (WSI) offer a way to capture fine needle aspiration smears and utilize image analysis tools to objectively evaluate morphologic features. Indeterminate classification of cases presents a challenge in cytopathology. The application of WSI image analysis in cytopathology will be detailed and potential uses discussed.

5:35 Close of Conference Program

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Cambridge Healthtech Institute's Second Annual

COMPANION DIAGNOSTICS

Implementing the New Standard

MONDAY, FEBRUARY 10

10:30 am Conference Program Registration

11:50 Chairperson's Opening Remarks

Julien N. Bradley, MBA, Senior Director, Sales & Marketing, Quanterix Corporation

» 12:00 pm KEYNOTE PANEL DISCUSSION:

CPT Coding

Co-Organized with AMP Association for Molecular Pathology

Moderator: Jill Hagenkord, M.D., CMO & Senior Vice President, InVitae (2013 AMP Economic Affairs Committee and 2014 AMP Training and Education Committee)

Elaine Lyon, Ph.D., Medical Director, Molecular Genetics, ARUP (2014 AMP President and 2013 AMP Economic Affairs, Professional Relations, Strategic Opportunities Committees)

Chris L. Jagmin, M.D., Senior Medical Director, National Medical Policy and Operations, Aetna

- Overview of CPT codes
- Transitioning to the molecular pathology codes from the clinical laboratory's perspective
- Understanding rationale behind new CPT codes

1:00 Session Break

1:15 Luncheon Presentation I: Simoa HD-1: A Fully Automated, Multiplexed Immunoanalyzer with Single Molecule Sensitivity

Sponsored by Quanterix

David C. Duffy, Ph.D., CTO, Quanterix Corporation

Single Molecule Array (Simoa) technology allows multiple proteins to be detected at concentrations 1000-fold lower than currently possible. Simoa is based on the capture of single molecules on paramagnetic beads, and their detection in arrays of femtoliter wells. We will illustrate the power of this analytical sensitivity in diagnosing cancer, neurological diseases, and infectious diseases using the Simoa HD-1 Analyzer, a fully automated instrument designed for use in clinical research and diagnosis.

1:45 Luncheon Presentation II A Novel Exosome RNA Extraction Platform Enabling Biomarker Discovery and Diagnostic Development for Personalized Medicine

Sponsored by exosome diagnostics

Johan Skog, Ph.D., CSO, Exosome Diagnostics

Exosomes are released by cells as an active process of communication, and contain stable, intact nucleic acids, making them an ideal source for biomarker discovery and diagnostic development. ExoRNeasy was optimized to extract RNA, including mRNA, microRNAs, and other RNAs, from plasma and serum. The high quality RNA generated from the ExoRNeasy kit enables profiling of tumor associated mutations as well as RNA levels in biofluids of cancer patients.

2:15 Session Break

2:30 PANEL DISCUSSION: Opportunities and Challenges in Commercializing Esoteric Diagnostics

Co-Organized with PMC PERSONALIZED MEDICINE COALITION

Moderator: Edward Abrahams, Ph.D., President, Personalized Medicine Coalition

Organized by the Personalized Medicine Coalition, this panel will outline both the promise and the pitfalls in bringing new molecular diagnostics to market, including the scientific, regulatory, payment, and adoption issues that innovative diagnostics companies must negotiate. Featuring leaders in the field, it will provide insight into best practices and likely scenarios for future trends.

Laura Brege, MBA, CEO, Nodality, Inc.

Peter Maag, Ph.D., CEO, XDx, Inc.

Jay Wohlgemuth, M.D., Senior Vice President, Science & Innovation, Quest Diagnostics

Alan Wright, M.D., MPH, CMO, Roche Diagnostics

3:45 Next-Gen Sequencing for Prostate Cancer

Sponsored by ResearchDX Your Companion in Diagnostics

Philip D. Cotter, Ph.D., FACMG, FFSc(RCPA), Principal, ResearchDx

Diagnostic and prognostic assessment of prostate cancer has been dramatically improved by the use of new laboratory technologies. This presentation focuses on the use of gene panel and pathway analyses using Next-Gen sequencing approaches in prostate cancer.

4:00 Models for the Development of Multiplex Companion Diagnostics

Sponsored by ALMAC

Austin Tanney, Ph.D., Scientific Liaison Manager, Almac

4:15 A Bioinformatics Framework for Clinical Research Sequencing

Sponsored by life technologies™

Daniel Rhodes, Ph.D., Head, Medical Science Informatics, Life Technologies

The widespread adoption of precision medicine in oncology requires: a compendium of therapies targeting the genetic vulnerabilities of cancer; the diagnostic tools capable of generating a precise molecular diagnosis; and importantly, the information systems to connect a patient's molecular diagnosis to optimal treatment hypotheses. Here, we will present Life Technologies' work to accelerate (1) defining the landscape of actionable driver genetic events in cancer, (2) developing a next-generation sequencing screen on the Ion Torrent semiconductor-sequencing platform, (3) assembling treatment-related information relevant for clinical research, and (4) devising a turnkey bioinformatics solution to democratize clinical sequencing in the future.

4:30 Refreshment Break and Transition to Plenary Keynote

» 5:00 PLENARY KEYNOTE SESSION
(please see page 4 for details)

6:15 Grand Opening Reception in the Exhibit Hall with Poster Viewing

7:45 Close of Day

TUESDAY, FEBRUARY 11

7:00 am Registration and Morning Coffee

» 8:00 PLENARY KEYNOTE SESSION
(please see page 4 for details)

9:15 Refreshment Break in the Exhibit Hall with Poster Viewing

Sponsored by

10:25 REGULATORY PANEL DISCUSSION: Late-Breaking News from the FDA that Impacts *in vitro* Diagnostic Tests

Moderator: Thomas F. Soriano, President & CEO, DOCRO, Inc.

This session will feature guest speakers from the FDA and industry who will discuss late-breaking news related to companion diagnostics (CDx), next-generation sequencing, (NGS), laboratory-developed tests (LDTs), including guidance documents. Coordination of the pharmaceutical and diagnostic components for approval will be discussed - questions having to do with the process for approval of drugs that require a CDx, what to do with the various data, and how to anticipate requirements during the clinical stages of development will be discussed.

Guest Speaker via Webex: Alberto Gutierrez, Ph.D., Director, Office of in Vitro Diagnostics (OIR), FDA

Richard Naples, Senior Vice President, Worldwide Corporate Regulatory Affairs, Becton-Dickinson

Michael Page, Ph.D., Senior Director, Oncology, Global Regulatory Affairs, Eisai, Inc.

Daniel J. O'Shannessy, Ph.D., Senior Director, Translational Medicine & Diagnostics, Morphotek

12:00 pm Optimizing Evidence Development and

COMPANION DIAGNOSTICS
CONTINUED ON NEXT PAGE

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COMPANION DIAGNOSTICS

continued

Communication to Support Market Access for Molecular Diagnostics
Joseph V. Ferrara, President, Boston Healthcare

12:30 Session Break

12:40 Luncheon Presentation I: Personalized Patient Therapy with Pharmacogenetics
Sponsored by Luminex
Stuart A. Scott, Ph.D., Assistant Professor, Genetics and Genomic Sciences, Icahn School of Medicine at Mount Sinai

1:10 Luncheon Presentation II Washington Legal Update – Strategies for Navigating Key Reimbursement and FDA Issues for Diagnostics
Sponsored by Sidley Austin LLP
Torrey Cope, J.D., Partner, FDA Regulatory, Sidley Austin LLP
Barbara Cammarata, J.D., M.P.H., Counsel, Health Care Regulatory/ Reimbursement, Sidley Austin LLP
Dora L Hughes, M.D., M.P.H., Senior Policy Advisor, Government Strategies, Sidley Austin LLP
This session will feature an FDA lawyer, a health care reimbursement/CLIA/fraud and abuse lawyer, and a government strategies expert who will discuss some of the key legal and policy issues facing diagnostics companies in the current regulatory environment. The panel will explain practical strategies companies may employ to address legal challenges related to commercializing and marketing their products under FDA and health care requirements, as well as strategies to obtain appropriate reimbursement and to comply with government and private payor reimbursement requirements. It will also cover the industry's role in developing law and policy to address the unique challenges in the emerging areas of *in vitro* diagnostics, companion diagnostics, molecular pathology and next generation sequencing.

1:40 Refreshment Break in the Exhibit Hall with Poster Viewing

2:15 STRATEGIC PANEL DISCUSSION: Recent Companion Diagnostic Drug Approvals
Moderator: Hakan Sakul, Ph.D., Executive Director and Head, Diagnostics, Worldwide R&D, Clinical Research and Precision Medicine; La Jolla Site Head, Development Operations, Pfizer, Inc.
Overview on diagnostic commercialization, reading market forces and what it takes to make a success.
Ron Mazumder, Ph.D., MBA, Global Head, Research & Product Development, Janssen/J&J
Peter Collins, Ph.D., Vice President & Head, Diagnostics, GlaxoSmithKline
Jeremy Bridge-Cook, Ph.D., Senior Vice President, Research & Development, Luminex
Eric Lai, Ph.D., Senior Vice President & Head, Pharmacogenomics, Takeda Pharmaceuticals
Gregory Zdechlik, COO, Eli Lilly & Co.

3:50 Sposnored Presentation
Sponsored by MRIGlobal
Speaker to be Announced

4:20 Late Breaking Presentation

4:35 Valentine's Day Celebration in the Exhibit Hall with Poster Viewing

5:20 Breakout Discussions in the Exhibit Hall *(see website for details)*

6:30 Close of Day

WEDNESDAY, FEBRUARY 12

7:00 am Breakfast Presentation *(Sponsorship Opportunity Available)* **or Morning Coffee**

» 8:00 PLENARY KEYNOTE SESSION
(please see page 4 for details)

9:45 Refreshment Break and Poster Competition Winner Announced in the Exhibit Hall

BUILDING THE CASE FOR REIMBURSEMENT- PRACTICAL CASE STUDIES

10:35 Chairperson's Remarks
Alan B. Carter, President, MDx Consulting

10:40 ConfirmMDx for Prostate Cancer: Navigating a Dynamic and Challenging Reimbursement Landscape
Jan Groen, Ph.D., CEO, MDx Health
Molecular diagnostics have lead to a significant improvement in diagnosing, treating and managing patient care. Despite significant advancements, labs and MDx innovators are faced with challenges encompassing coding, billing, and attempting to obtain coverage and fair reimbursement. The evidence bar is rising, with the burden on the MDx laboratory to prove clinical utility. In this case study we discuss the challenging reimbursement path for a new epigenetic test for prostate cancer.

11:10 Found in Translation: Taking the Cardiac Biomarker ST2 from Discovery to Clinical Adoption
James V. Snider, Ph.D., President, Critical Diagnostics
This presentation will touch on the discovery research and original identification ST2 as a potential cardiac biomarker. The emphasis though will be on the effort to develop a validated assay and the clinical evidence to justify routine clinical use of ST2 as a cardiac disease patient management tool, and will conclude with a discussion of the regulatory and reimbursement landscape.

11:40 Better Clinical Outcomes through Cardiovascular Genomics
Lon Castle, M.D., Medical Science Liaison, Medical Affairs, CardioDx
As the upcoming tidal wave of genomic tests floods the market, the molecular diagnostic industry will be faced with convincing payers that a new test provides enough value to warrant reimbursement. This presentation will focus on how a company can navigate the evidence development process in order to sail through a payer's technology assessment committee. The importance of testing the waters with an internal economic forecast modeling the impact of coverage will also be discussed.

12:10 pm Optimize for Variant Calling Confidence: Impact of Sample Type and Analytical Methods on NGS Data Quality
Sponsored by DNA genotek
Milos Popovic, Technical Product Manager, Seven Bridges Genomics
Seven Bridges Genomics, in collaboration with DNA Genotek, will present data and best practises for variant calling based on whole genome sequencing of DNA from matched pairs of saliva collected and stabilized with Oragene vs blood. When introducing personalized medicine offerings it is imperative to consider all parts of the patient value chain from sample collection to processing and validated results. Optimizing the patient experience, ensuring quality samples and future-proofing lab analysis through high quality specimen inputs is key to enabling successful, scalable health solutions.

12:25 Luncheon Presentation I:
Sponsored by Precision for Medicine
David Parker, Ph.D., Vice President, Market Access Strategy, Precision for Medicine
Judi Smith, Vice President, in vitro Diagnostics and Quality, Precision for Medicine

1:00 Refreshment Break in the Exhibit Hall and Last Chance for Poster Viewing

CHANGING CLINICAL DECISION SUPPORT BASED ON MOLECULAR DIAGNOSTICS

1:40 Chairperson's Remarks
Harry Glorikian, Managing Director, Strategy, Precision for Medicine

» 1:45 KEYNOTE PRESENTATION:

New Frontiers in Medical Diagnostics
Carlos Cordon-Cardo, M.D., Ph.D., Professor & Chair Pathology; Professor Genetics and Genomic Sciences; Professor Oncological Sciences, Mount Sinai Hospital

2:15 EXPERT PANEL DISCUSSION
With the dramatic increase in data being generated by molecular diagnostic devices (NGS in particular), very few clinical decisions are "binary" in nature today. Further, new technologies and clinical findings (mutations, pathways, drug resistance profiles) are being introduced regularly. To help compensate for the complexity and data overload, clinicians are hopeful that clinical decision support systems will meet the challenge. The panel will discuss their aspirations for these systems and whether existing solutions are solving their needs.
Panelists: Carlos Cordon-Cardo, M.D., Ph.D., Professor & Chair Pathology; Professor Genetics and Genomic Sciences; Professor

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COMPANION DIAGNOSTICS

continued

Oncological Sciences, Mount Sinai Hospital
Kathryn A. Teng, M.D., FACP, Director, Center for Personalized Healthcare, Staff Physician, Internal Medicine, Cleveland Clinic; Assistant Professor, CWRU Lerner College of Medicine
Christopher D. Gocke, M.D., Associate Professor of Pathology and Oncology, Director, Heme Molecular Diagnostics, Johns Hopkins University School of Medicine

EU REGULATORY KEYNOTE

3:15 Chairperson's Remarks
Richard A. Montagna, Ph.D., Senior Vice President, Scientific Affairs, Rheonix, Inc.

3:20 Diagnostics Regulation and Policy in the EU
Hans-Georg Eichler, M.D., Senior Medical Officer, European Medicines Agency (EMA)

3:45 Refreshment Break

BIG DATA DRIVING PERSONALIZED MEDICINE

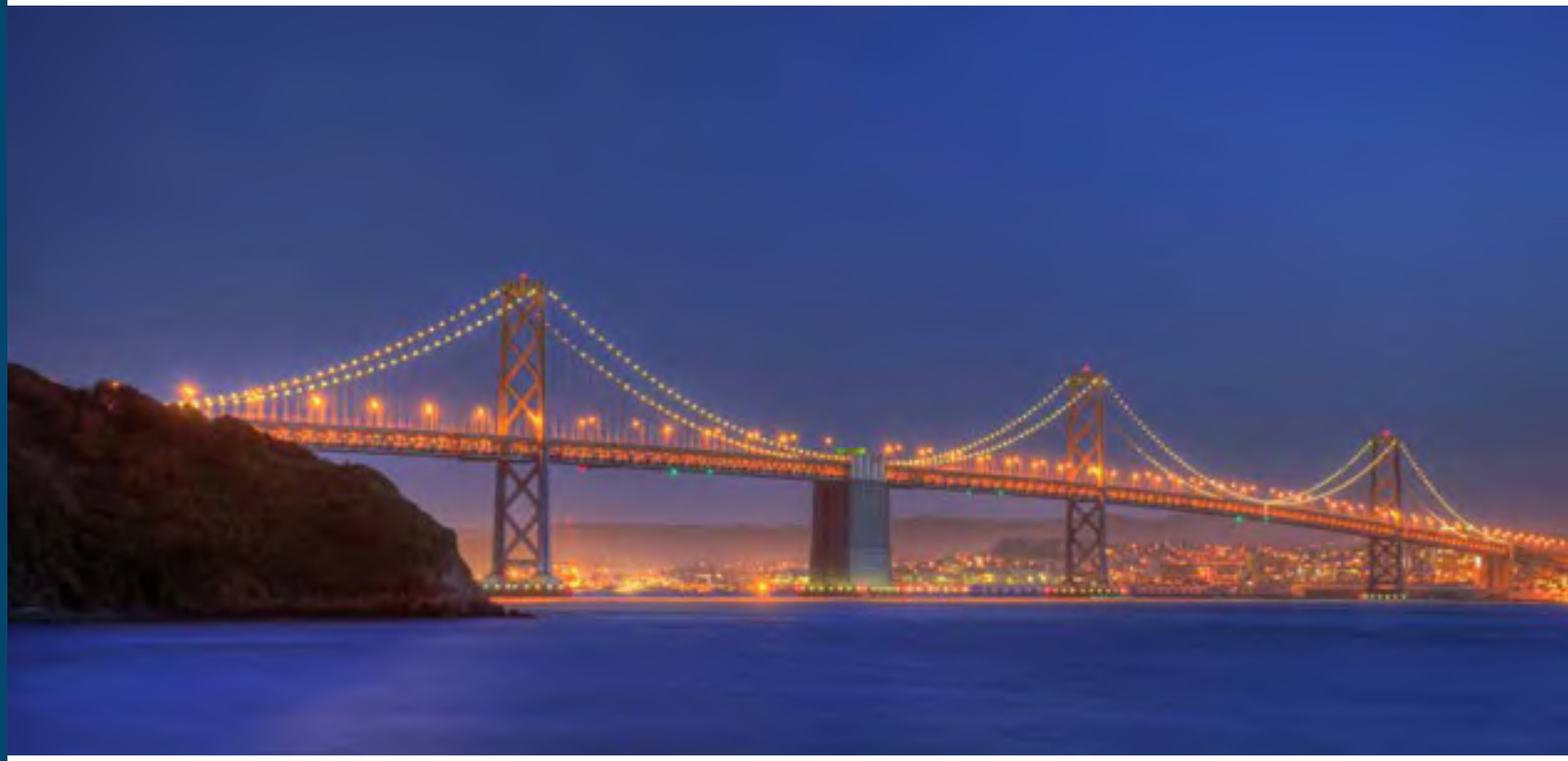
4:00 Chairperson's Remarks
Dalia Cohen, Ph.D., Founder & President, ALN Associates

4:05 Big Data's Big Role in Understanding Complex Diseases
Andreas Kogelnik, M.D., Ph.D., Founder and Director, Open Medicine Institute
The Open Medicine Institute (OMI) is effectively applying a collaborative, "big data" approach to understand and address complex diseases including: Autism, Lyme, Chronic Fatigue Syndrome, Parkinson's, and various cancers. This presentation will discuss the creation of a patient-centric infrastructure that handles and analyzes genomic sequencing information, bio-sampling data, physiology tests, basic research, patient experiences and physician evaluations to deliver needed information about a range of diseases.

4:35 Progress on Aggregating all the World's Genetic Tests into a Single Assay
Randy Scott, Ph.D., CEO and Co-Founder, InVitae
Technology is moving rapidly to enable massively parallel genetic testing. The ability to sequence DNA, however, is only the first step in building the infrastructure to analyze, store, manage, and interpret medical genetic information for patients. InVitae is focused on building the infrastructure to bring more comprehensive genetic testing into routine medical practice throughout the world.

5:05 It's Not Just About Big Data...Big Analytics for Identifying What Works and for Whom in Healthcare
Iya Khalil, Ph.D., Executive Vice President and Co-Founder, GNS Healthcare
We are living in the era of big data in healthcare, with unprecedented ability to collect data at multiple levels (genomic/'omic', phenotypic, health records, mobile health, etc.) and at scale. The key will be leveraging advanced analytics and appropriate feedback loops to identify what works on an individual patient level.

5:35 Close of Conference Program



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Cambridge Healthtech Institute's Inaugural

PCR FOR MOLECULAR MEDICINE

Current Applications and Future Perspectives

MONDAY, FEBRUARY 10

10:30 am Conference Program Registration

» KEYNOTE SESSION

11:50 Chairperson's Opening Remarks
Carl Wittwer, M.D., Ph.D., Professor, Pathology, University of Utah

12:00 pm SuperSelective Primers for the Detection of Rare Mutant DNA from Cancer Cells in Samples Containing Abundant Wild-Type DNA
Fred Russell Kramer, Ph.D., Professor, Department of Microbiology and Molecular Genetics, Public Health Research Institute, New Jersey Medical School
"SuperSelective" primers, by virtue of their unique design, enable only a few molecules of a mutant sequence to generate amplicons in conventional, real-time PCR assays without interference from extremely abundant wild-type molecules, even if the only difference between the mutant sequence and the wild-type sequence is a single-nucleotide polymorphism. As few as 10 mutant molecules can routinely be distinguished and quantitated in samples containing 1,000,000 wild-type molecules.

12:30 Digital PCR Goes COLD: Application of COLD-PCR in Digital Format Enables Quantitative and Multiplexed Mutation Scanning
G. Mike Makrigiorgos, Ph.D., Professor, Radiation Oncology, Dana Farber, Harvard Medical School
As currently applied, digital PCR can only be used to detect known mutations at single sequence positions. By merging COLD-PCR and digital-PCR technologies we enable digital mutation scanning, a new frontier for digital PCR. A single reaction can now interrogate numerous mutations, and replaces multiple individual digital PCR reactions.

1:00 Session Break

1:15 Luncheon Presentations *(Sponsorship Opportunities Available)* **or Lunch on Your Own**

2:15 Session Break

IMPROVEMENTS TO QPCR

2:30 Chairperson's Remarks
Adam R. Abate, Ph.D., Assistant Professor, Bioengineering and Therapeutic Sciences, California Institute for Quantitative Biosciences (QB3), University of California, San Francisco

2:35 Extreme PCR: Efficient and Specific Amplification in <1 Min by Selected Use of PCR Components
Carl Wittwer, M.D., Ph.D., Professor, Pathology, University of Utah
Although 10 min PCR was first reported in 1990, further decreases in time to result usually sacrifice efficiency and yield. Extreme PCR combines <2 s cycles with high primer and polymerase concentrations. Cycle times of <1 s for PCR products under 100 bp in length provide specific, high yield amplification. Effects of the type of polymerase, monovalent cations, Mg++, Tm depressors, and noncovalent dyes on polymerase extension rates are also reported using a new polymerase activity assay.

3:05 TINA Primers for Real-Time TaqMan PCR
Uffe Vest Schneider, M.D., Ph.D., CSO, QuantiBact A/S
TINA modified primers improve flexibility in primer design, PCR multiplexing capacity and assay robustness in PCR assays. We here present data from a tetraplex TaqMan PCR assay targeting methicillin-resistant Staphylococcus aureus, demonstrating significantly better assay efficacy and analytical sensitivity by TINA modified primers compared to unmodified DNA primers.

3:35 Virtual Barcoding Using LATE-PCR and Its Allied Technologies
Lawrence J. Wangh, Ph.D., Professor, Biology, Brandeis University
LATE-PCR and its allied technologies make it possible to accurately amplify multiple single-stranded products and then analyze these products at end-point in multiple colors over a wide range of temperatures. The Barcode of Life Project focuses on the COX1 gene of mitochondria to uniquely identify all animals on earth. We are developing Virtual Barcoding, a rapid, reliable closed-tube method for species identification anywhere on earth.

4:05 FEATURED POSTER PRESENTATION: Optical Control and Thermocycling Calibration of Laser-Heated Microdroplet PCR
Eric Hall, Ph.D., Postdoctoral Fellow, Molecular Physics, SRI International
Aqueous droplets containing lysis and polymerase chain reaction (PCR) reagents are deposited on top of single cells under a layer of oil. Following extraction of their genetic content, PCR and reverse transcription PCR (RT-PCR) may be performed on cells of interest in a rapid fashion via infrared-laser heating. Microdroplets may be deposited on a variety of substrates, requiring only a change in the lecithin content of the oil phase to produce droplets with the correct, spherical geometry. The thermocycling protocol employed in laser-heated PCR may be calibrated via melting of two hairpin oligos, each labeled with a matched reporter-quencher pair. The fluorescent labels on the hairpin oligos are spectrally distinct from those on the primer probes used in PCR, allowing for each droplet to be automatically calibrated during the initial heating ramp to the hot activation of polymerase, controlling for heating variations that may arise from changes in droplet shape and position.

4:35 Refreshment Break and Transition to Plenary Keynote

» 5:00 PLENARY KEYNOTE SESSION
(please see page 4 for details)

6:15 Grand Opening Reception in the Exhibit Hall with Poster Viewing

7:45 Close of Day

TUESDAY, FEBRUARY 11

7:00 am Registration and Morning Coffee

» 8:00 PLENARY KEYNOTE SESSION
(please see page 4 for details)

9:15 Refreshment Break in the Exhibit Hall with Poster Viewing

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NOVEL APPROACHES AND NEW WAYS OF THINKING

10:25 Chairperson's Remarks
Robert Palais, Ph.D., Associate Professor, Math Dept, Utah Valley University; Research Professor, Pathology Dept, University of Utah

PCR FOR MOLECULAR MEDICINE
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PCR FOR MOLECULAR MEDICINE

continued

10:30 Counting Copies: Digital Distributions and Math for Melting

Robert Palais, Ph.D., Research Professor, Mathematics; Professor, Pathology, University of Utah

We will discuss two mathematical algorithms that can be used to quantify DNA copy number variation with increased accuracy. One permits rapid exact evaluation of cumulative probability distributions having one or more degrees of freedom, for sampling with or without replacement. Another normalizes high-resolution melting peak of the target with respect to that of a reference after ensuring the initial copy ratio is preserved during PCR.

11:00 Eprobe: New Fluorescence Probe for the Combination of Real-Time PCR and Melting Curve Analysis

Takeshi Hanami, Ph.D., LSA Technology Development Unit, Omics Science Center, RIKEN

Eprobe is a hybridization-sensitive fluorescent probe that only shows strong fluorescence signals upon hybridization to a complementary DNA strand. This talk will describe how the probe combines real-time PCR monitoring and melting curve analysis as well as how we achieved multiplex detection by using multicolor Eprobes. The combination provides powerful means for new mutation detection assays in a single-tube reaction.

11:30 PACS: PCR-Activated Cell Sorting

Adam R. Abate, Ph.D., Assistant Professor, Bioengineering and Therapeutic Sciences, California Institute for Quantitative Biosciences (QB3), University of California, San Francisco

We have developed a microfluidic system that allows individual cells to be analyzed and sorted based on the outcomes of single-cell PCR reactions. Our system encapsulates each cell in a microdroplet, lyses the cell, and performs RT-PCR to detect gene transcripts, SNPs, or small non-coding RNAs of interest. Unlike FACS, PACS requires no antibodies and can differentiate among cells based on transcriptional variation. The technology has broad applications in basic research and medical diagnostics, including in cancer, immune function, and microbiology.

12:00 pm Selected Poster Presentation

12:30 Session Break

12:40 Luncheon Presentations *(Sponsorship Opportunities Available)* **or Lunch on Your Own**

1:40 Refreshment Break in the Exhibit Hall with Poster Viewing

THE RISE OF DIGITAL PCR

2:15 Chairperson's Remarks

Fred Russell Kramer, Ph.D., Professor, Department of Microbiology and Molecular Genetics, Public Health Research Institute, New Jersey Medical School

2:20 Application of Digital PCR for the Analysis of RNA

Rebecca Sanders, Researcher, Nucleic Acid Metrology, Molecular Biology, Science and Technology Division, LGC

2:50 Digital PCR Developments We'd Like to See

N. Reginald Beer, Ph.D., Medical Diagnostics Initiative Leader, Center for Micro and Nanotechnologies, Lawrence Livermore National Laboratory

The development of digital PCR has led to many exciting applications that benefit from the technique's ability to isolate, amplify, and detect rare or numerically-disadvantaged targets. Not surprisingly, this created early interest in medical diagnostics where the ability to detect rare mutants among a wild type background is critical. More widespread adoption, however, would benefit from additional advances that increase the technology's value proposition. In this talk we will discuss several trends that could broaden dPCR's appeal.

3:20 Picoliter Droplet-Based Digital PCR for Molecular Diagnostics

Valerie Taly, Ph.D., Group Leader/CNRS Researcher, Université Paris-Descartes

Picoliter droplet-based digital PCR allows the highly sensitive and quantitative detection of rare cancer markers within complex mixtures of DNA like patient samples. We will present the development of droplet multiplex procedures for the quantitative detection of the seven most frequent KRAS mutant alleles as well as wild type sequences and clinical applications of these procedures for patient treatment management. The results of two clinical trials involving metastatic cancer patients will be presented.

3:50 Liquid Biopsy – Blood-Based Molecular Testing in Oncology

Sponsored by sysmex

Frank Diehl, Ph.D., Vice President, Lab Operations, Research & Development, Sysmex Inostics

The use of blood for molecular testing in oncology opens up new, non-invasive possibilities for the management of cancer patients in the context of therapy selection, response prediction, real time follow-up, and resistance monitoring.

4:05 Sponsored Presentation *(Opportunity Available)*

4:20 Valentine's Day Celebration in the Exhibit Hall with Poster Viewing

5:20 Breakout Discussions in the Exhibit Hall *(see website for details)*

6:30 Close of Day

WEDNESDAY, FEBRUARY 12

7:00 am Breakfast Presentation *(Sponsorship Opportunity Available)* **or Morning Coffee**

» **8:00 PLENARY KEYNOTE SESSION**
(please see page 4 for details)

9:45 Refreshment Break and Poster Competition Winner Announced in the Exhibit Hall

10:35 Chairperson's Remarks

N. Reginald Beer, Ph.D., Medical Diagnostics Initiative Leader, Center for Micro and Nanotechnologies, Lawrence Livermore National Laboratory

10:40 PANEL DISCUSSION: Future Directions for PCR

Moderator: N. Reginald Beer, Ph.D., Medical Diagnostics Initiative Leader, Center for Micro and Nanotechnologies, Lawrence Livermore National Laboratory

Panelists: Christopher D. Gocke, M.D., Pathology; Director of Hematology Molecular Diagnostics and Program Director, Molecular Genetic Pathology Fellowship, Johns Hopkins University

Matthew Strain, M.D., Ph.D., Assistant Professor, Center for AIDS Research, University of California, San Diego

Bernhard Zimmermann, Ph.D., Senior Director, Research & Development, Natera, Inc.

APPLICATIONS IN PRENATAL DIAGNOSTICS

11:10 Noninvasive Prenatal Diagnosis through Genetic Molecular Analysis of Circulating Trophoblastic Cells

Patrizia Paterlini-Brechot, M.D., Ph.D., Professor, Cell Biology and Oncology, University Paris Descartes Paris; Director, INSERM

Trophoblastic cells are expected to provide the optimal DNA substrate for noninvasive prenatal diagnosis (NI-PND) as they carry fetal DNA not mixed with maternal DNA, and circulate at very early terms of pregnancy. Results of a clinical validation study concerning NI-PND of hereditary genetic diseases using circulating trophoblastic cells, and of its potential extension, will be discussed.

11:40 Massively Multiplexed Single-Nucleotide Polymorphism Amplification and Sequencing to Identify Fetal Aneuploidy from Cell-Free DNA in Maternal Circulation

PCR FOR MOLECULAR MEDICINE
CONTINUED ON NEXT PAGE

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continued

Bernhard Zimmermann, Ph.D., Senior Director, Research & Development, Natera, Inc.

We developed a method to multiplex, in a single reaction, tens of thousands of PCR assays targeting SNPs. Noninvasive prenatal testing based on analysis of cell-free DNA (cfDNA) is one of the most rapidly-expanding molecular diagnostics fields. I will present the most recent performance data from a commercialized version of this approach, a highly accurate noninvasive prenatal aneuploidy test. I will also discuss the potential of this approach to detect various microdeletion syndromes, which is the next major challenge in noninvasive prenatal testing.

12:10 pm Session Break

12:20 Luncheon Presentation *(Sponsorship Opportunity Available)* **or Lunch on Your Own**

1:00 Refreshment Break in the Exhibit Hall and Last Chance for Poster Viewing

APPLICATIONS IN CANCER

1:40 Chairperson's Remarks

1:45 Tandem Duplication PCR: A Novel Method of Detecting Minimal Residual Disease and Minor Clones in FLT3/ITD AML

Christopher D. Gocke, M.D., Pathology; Director of Hematology Molecular Diagnostics and Program Director, Molecular Genetic Pathology Fellowship, Johns Hopkins University

The TD-PCR was initially designed to confirm ITD mutation of an amplicon, which was undetectable by capillary electrophoresis and was incidentally isolated by a molecular fraction collecting tool. Subsequently, TD-PCR detected ITD mutation in 2 of 77 patients previously reported as negative for ITD mutation by a standard PCR assay. TD-PCR can also potentially be applied to monitor minimal residual disease with high analytic sensitivity in a portion of ITD-positive acute myeloid leukemia patients.

2:15 Prediction of Lung Cancer Histological Types by RT-qPCR Gene Expression in FFPE Specimens

Mark R. Miglarese, Ph.D., CSO, GeneCentric Diagnostics

Lung cancer histologic diagnosis is clinically relevant because there are histology-specific treatment indications and contraindications. Diagnosis by standard histopathological methods can be challenging due to a variety of tumor characteristics as evidenced by less-than-ideal agreement among pathologists reviewing the same specimens. We developed a 57-gene RT-qPCR expression predictor of lung cancer histology for use on FFPE specimens. Assay performance and comparison to interpretations from traditional pathology results will be discussed.

2:45 BEAMing and Droplet Digital PCR Analysis of Mutant IDH1 mRNA in Glioma Patient Serum and Cerebrospinal Fluid Extracellular Vesicles

Leonora Balaj, Ph.D., Researcher, Massachusetts General Hospital, Harvard Medical School

Here, we describe a novel approach that uses BEAMing droplet RT-PCR (EV-BEAMing), as well picodroplet digital PCR, to interrogate mRNA sequences contained within EVs from serum and CSF of glioma patients. Using both assays, we were able to reliably detect and quantify mutant and wild-type IDH1 transcripts in CSF of patients with gliomas. EV-BEAMing and picodroplet dPCR from EV's represent a valuable new strategy for cancer diagnostics, which can be applied to a variety of biofluids and neoplasms.

3:15 Sponsored Presentations *(Opportunities Available)*

3:45 Refreshment Break

APPLICATIONS IN INFECTIOUS DISEASE

4:00 Chairperson's Remarks

Niaz Banaei, M.D., Assistant Professor, Pathology & Infectious Diseases, Stanford University

4:05 Applications of Digital PCR to Clinical Viral Load Testing

Randall T. Hayden, M.D., Director, Clinical and Molecular Microbiology, Member, Department of Pathology, St. Jude Children's Research Hospital

Viral load testing has become integral to the care of many critically ill patients, but currently available tests suffer from marked intra-laboratory result variability and a lack of standardization. Digital PCR offers a means of direct quantitative measurement that may improve both accuracy and precision compared to other methods. This session will describe the theory behind digital PCR, as well as some initial studies evaluating its use for viral load testing.

4:35 Application of Droplet Digital PCR for the Investigation of the Latent HIV Reservoir

Matthew Strain, M.D., Ph.D., Assistant Professor, Center for AIDS Research, University of California, San Diego

The latent reservoir of HIV represents the obstacle to curing the infection with the currently highly effective antiretroviral therapy. Measuring the reservoir requires assays of rare events in a large number of cells. We have developed and applied droplet digital PCR to the assay of various species of HIV DNA and RNA transcripts. The characterization of these assays and their application to clinical studies will be discussed.

5:05 Sequencing Comes to Rescue When All Else Fails: Pathogen Detection and Identification in Clinical Samples with Broad Range PCR Amplicon Sequencing

Niaz Banaei, M.D., Assistant Professor, Pathology & Infectious Diseases, Stanford University

Clinicians frequently encounter patients with life threatening culture-negative infections. Broad range PCR combined with amplicon sequencing is a powerful tool for rapid and accurate diagnosis of culture-negative or uncultured invasive infections. In this case presentation I will describe and discuss the utility and potential pitfalls of ribosomal RNA locus sequencing for direct detection and identification of invasive bacteria and fungi from fresh and formalin-fixed, paraffin-embedded specimens.

5:35 Close of Conference Program

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Cambridge Healthtech Institute's Inaugural

BIOSPECIMEN SCIENCE AND SAMPLE PREP

Enabling Sample-Centered Precision Medicine

MONDAY, FEBRUARY 10

10:30 am Conference Program Registration

ENABLING SAMPLE-CENTERED PRECISION MEDICINE

11:50 Chairperson's Opening Remarks

David L. Rimm, M.D., Ph.D., Professor, Pathology; Executive Director, Translational Pathology; Director, Yale Pathology Tissue Services, Yale University

12:00 pm BioSpecimen Sciences: Pioneering Sample-Centered Precision Medicine

Michael H.A. Roehrl, M.D., Ph.D., Director, UHN Program in BioSpecimen Sciences, University of Toronto

In the age of personalized molecular medicine, BioSpecimen Sciences and BioBanking are rapidly becoming the key drivers of success. We will explore important considerations related to patient sample quality, procurement, and database annotation for use in deep omic personalized molecular profiling for individualized therapy guidance, biomarker surveillance, and drug discovery. We will show examples of how BioSpecimen Sciences done right will enable a new generation of molecularly-driven clinical trials for drug development.

12:30 Genentech's Human Tissue Laboratory: Supporting Innovation in Translational and Basic Research within a Large and Rapidly Evolving R&D Setting

Cary D. Austin, M.D., Ph.D., Pathologist, Department of Pathology, Genentech, Inc.

Genentech's Human Tissue Laboratory supports a variety of research and development activities, including projects focused on drug and diagnostics development as well as those in basic research in the fields of oncology, immunology, neuroscience, and metabolism. Supporting such a diverse set of needs in a rapidly evolving organizational and industrial setting presents challenges that will be discussed.

1:00 Session Break

1:15 Luncheon Presentation I: Optimize Use: Increase Biospecimen Utility with Real-Time Annotation & Data Reconciliation of Clinical, Specimen and Ethical Attributes

Sponsored by PRECISION BIOSERVICES

Katheryn Shea, Vice President, Bioservices, Precision Bioservices

The foremost concern in specimen utilization is the reliability of the data associated with the specimens. Careful documentation of all use parameters including medical history, clinical manifestations, permitted and restricted sample uses per the informed consent, and physical attributes of the specimens are needed to determine if samples will be fit for purpose. Real-time, traceable data reconciliation processes ensure the quality of the data and usability of the specimens.

1:45 Luncheon Presentation II
(Sponsorship Opportunity Available)

2:15 Session Break

TISSUE SAMPLE QUALITY ASSESSMENT AND ASSURANCE

2:30 Chairperson's Remarks

Katheryn Shea, Vice President, Bioservices, Precision Bioservices, Inc.

2:35 General Pre-Analytical Issues in Clinical Diagnostics and Molecular Pathology

Gianni Bussolati, M.D., Professor, Department of Medical Science, University of Torino

Cold ischemia time, transfer and temperature and time to and of fixation constitute critical and potentially detrimental conditions for tissue preservation. Standardization of such variables will be approached with implementation of new technology and multidisciplinary expertises.

3:05 The Tissue Quality Index: A Denominator for Immunohistochemistry

David L. Rimm, M.D., Ph.D., Professor, Pathology; Executive Director, Translational Pathology; Director, Yale Pathology Tissue Services, Yale University

Standardization of immunohistochemistry (IHC) has been challenging, to the point that some scientists consider the assay inherently non-quantitative. In comparison, RNA measurements can be quantitative by using a series of housekeeping genes as a normalization factor that adjusts for the myriad and undefinable pre-analytic variables. Here we introduce the first steps toward providing a similar "denominator" for IHC. While not yet a true normalization factor, the tissue quality index has the potential to provide information on tissue integrity for biomarker assessment.

3:35 High Content Single-Cell Diagnostics in Oncology

Peter Kuhn, Ph.D., Associate Professor of Cell Biology, The Scripps Research Institute

Tumor heterogeneity is the result of both genomic instabilities and mircoenvironmental adaptations under both natural evolution of the disease and treatment pressures. Heterogeneity is mostly evaluated at the cellular level considering the individual cell as the biological unit. We have established a framework of single cell analyses that can integrated high content data at the phenotypic and genotypic level. The number of biological units/single cells analyzed provides the measure of resolution of the quantified heterogeneity. The high-content analysis utilizes the high-definition circulating tumor cell (HD-CTC) assay, which provides for an enrichment-free approach to identify and characterize CTCs. We utilized the HD-CTC assay to study protein biomarker expression combined with single-nucleus sequencing for genome-wide analysis of copy number variation (CNV) in fluid and solid biopsies with sequential sampling over the course of disease evolution. Standardized sample preparation methods that enables quantitative comparisons of multiple specimen types both intra- and inter-patient as well as along the timeline of cancer evolution. This presentation will focus on both pre-analytical validation and research results in multiple cancer types.

4:05 Improving Upstream Sample Management and Downstream Bioprocessing

Sponsored by BioStorage Technologies

Kristina Robson, Ph.D., Senior Director, Comprehensive Solutions, BioStorage Technologies

Robust sample inventory and bioinformatics systems are fundamental to optimizing the value of biological sample collections. This session will cover our BioProcessing Solutions integration of a streamlined process for sample management and downstream bioprocessing.

4:20 Orthogonal PCR: Sample to Actionable Results Using Coupled qPCR and High Multiplex Endpoint Detection

Sponsored by Rheonix

Richard A. Montagna, Ph.D., Senior Vice President, Scientific Affairs, Rheonix, Inc.

The limitations of current real time PCR systems (qPCR) for multiplexing have been overcome by combining four color qPCR with simultaneous detection of 50 additional targets on a DNA array. The orthogonal system allows raw specimens to be introduced and automatically processed through sample preparation, analysis and readout without any user intervention.

BIOSPECIMEN SCIENCE AND SAMPLE PREP
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BIOSPECIMEN SCIENCE AND SAMPLE PREP

continued

4:35 Refreshment Break and Transition to Plenary Keynote

» 5:00 PLENARY KEYNOTE SESSION
(please see page 4 for details)

6:15 Grand Opening Reception in the Exhibit Hall with Poster Viewing

7:45 Close of Day

TUESDAY, FEBRUARY 11

7:00 am Registration and Morning Coffee

» 8:00 PLENARY KEYNOTE SESSION
(please see page 4 for details)

9:15 Refreshment Break in the Exhibit Hall with Poster Viewing

Sponsored by

SAMPLE PREP CONSIDERATIONS FOR NGS-BASED CLINICAL ASSAYS

10:25 Chairperson's Remarks

Martin Siaw, Ph.D., Associate Scientific Director, Advanced Sequencing, Quest Diagnostics Nichols Institute

10:30 NGS-Based Clinical Assays: Building Castles in the Air

Jamie L. Platt, Ph.D., Scientific Director, Advanced Sequencing, Quest Diagnostics Nichols Institute

While applying NGS in the clinical setting may seem like "building castles in the air" to some, the utility of NGS assays can be enormous when built on a strong foundation. The foundation of sample prep will readily prove the validity of "garbage in, garbage out". Examples of sample prep issues from commercially available NGS tests will be introduced.

11:00 Optimized Sample Handling and Target Enrichment Strategies for Challenging Clinical Samples

Bill Biggs, Ph.D., Director, Clinical Sequencing, Broad Institute of MIT and Harvard

The desire to access valuable clinical FFPE samples using advanced molecular techniques such as next-generation sequencing methods in an efficient and productive manner represents an ongoing challenge for most clinical laboratories. At the Clinical Research Sequencing Platform (CRSP) within the Broad Institute methods have been developed, optimized and implemented which allow for the ready and routine access of FFPE samples for such NGS-based analyses as Whole Exome Sequencing and Targeted Re-sequencing. These processes and methods allow for exceptional data to be obtained from even the most challenging of FFPE samples where low yield (≤ 100ng) or age (> 5 yrs) can confound analytical processes.

11:30 Automation of Sample Preparation for Clinical NGS: The Requirements and the Challenges Presented by the Various Clinical Sample Types

Martin Siaw, Ph.D., Associate Scientific Director, Advanced Sequencing, Quest Diagnostics Nichols Institute

Sample preparation is an important component of any molecular testing that is being done in clinical laboratories. With the increasing use of NGS for clinical testing comes the need to process increasingly larger numbers of patient samples. Automation of sample preparation should be considered to be critical to the workflow of diagnostic tests involving the use of NGS. My presentation will focus on the requirements and challenges for CLIA-certified clinical laboratories

12:00 pm Better Annotation Needed at Various Points in the Sequencing Process to Avoid Reporting Errors due to Pseudogenes and High Repeat Genomic Regions

Patricia Mueller, Ph.D., Chief, Molecular Risk Assessment Laboratory, Newborn Screening and Molecular Biology Branch, DLS, Centers for Disease Control and Prevention (CDC)

Outside of clinical genomic sequencing centers, there is a general lack of awareness of the problems in NGS caused by the large number of pseudogenes in the human genome. Current estimates exceed 19,000 pseudogenes. Current standard enrichment methods do not reliably distinguish between highly homologous functional and pseudogenes. Better annotation is needed at different points in the sequencing process, including reference sequences, hybrid-capture arrays, PCR primer design software, and data-analysis software to help those designing tests and analyzing data to avoid reporting errors.

12:30 Session Break

12:40 Luncheon Presentation I: Seven Steps to the Sample Life: Best Practices for Clinical Trial Sample Management

Sponsored by BioFortis

Mark. A Collins, Ph.D., Director, Marketing, BioFortis, Inc.

The increased interest in biomarker-based studies necessitates a new rigor and sophistication in sample and sample related data management within the clinical trial context. In addition many trials occur across geographies, are increasingly externalized with multiple stakeholders and generate large amounts of data, which is putting existing software systems, infrastructures and processes under considerable pressure. Using case studies, attendees will learn emerging best practices for clinical trial sample and data management.

1:10 Luncheon Presentation II *(Sponsorship Opportunity Available)*

1:40 Refreshment Break in the Exhibit Hall with Poster Viewing

SPECIMEN CONSIDERATIONS IN BIOMARKER-DRIVEN CLINICAL TRIALS

2:15 Chairperson's Remarks

Michael H.A. Roehrl, M.D., Ph.D., Director, UHN Program in BioSpecimen Sciences, University of Toronto

2:20 Use of Clinical Genetic Specimens to Support Clinical Drug Development

Peter Shaw, Ph.D., Senior Principle Scientist, Clinical Pharmacogenomics, Merck & Co., Inc.

Many pharmaceutical companies maintain biorepositories of clinical specimens consented for and collected during clinical trials. This presentation will address common uses of biorepository samples to support clinical drug development, and will highlight challenges in specimen collection that impact ultimate utility of the specimen.

2:50 Biospecimen Sample Integrity and Validation of Biomarkers in Clinical Trials

Lokesh Agrawal, Ph.D., Program Director, Biorepositories and Biospecimen Research Branch (BBRB), Cancer Diagnosis Program, Division of Cancer Treatment and Diagnosis, National Cancer Institute

3:20 Best Practices for Management of Clinical Specimens to Support Clinical Drug Development

George Y. Tokiwa, Ph.D., Associate Principal Scientist, Clinical Genetics, Merck Sharp & Dohme Corp.

Many pharmaceutical companies maintain biorepositories of clinical specimens collected during clinical studies that are consented for future biomedical research. This presentation will focus on best practices to address the end to end clinical specimen management activities required to support optimal and compliant use of the specimens.

3:50 When Worlds Collide: Sharing A Vision for an Integrated Clinical Trial Management System

Sponsored by REMEDY INFORMATICS

Bruce Pharr, Vice President, Product Marketing Laboratory Systems, Remedy Informatics

Mr. Pharr will discuss data strategies to facilitate a more efficient flow of information enterprise-wide between disparate, yet key players in life sciences research and healthcare—such as clinical trial teams, hospitals, labs, biobanks, and others—in order to deliver on and advance the promise of personalized medicine.

BIOSPECIMEN SCIENCE AND SAMPLE PREP
CONTINUED ON NEXT PAGE

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continued

4:20 Valentine's Day Celebration in the Exhibit Hall with Poster Viewing

5:20 Breakout Discussions in the Exhibit Hall *(see website for details)*

6:30 Close of Day

WEDNESDAY, FEBRUARY 12

7:00 am Breakfast Presentation *(Sponsorship Opportunity Available)* **or Morning Coffee**

» **8:00 PLENARY KEYNOTE SESSION**
(please see page 4 for details)

9:45 Refreshment Break and Poster Competition Winner Announced in the Exhibit Hall

SAMPLE QUALITY ISSUES IN PROFICIENCY TESTING AND CERTIFICATION

10:35 Chairperson's Remarks
Nazneen Aziz, Ph.D., Director, Molecular Medicine, Transformation Program Office, College of American Pathologists

10:40 College of American Pathologists/CLIA Standards for Next-Generation Sequencing and Proficiency Testing
Nazneen Aziz, Ph.D., Director, Molecular Medicine, Transformation Program Office, College of American Pathologists
Within 5 years of its introduction and widespread use in research, NGS is transforming molecular medicine. NGS has been adopted into clinical testing far more rapidly than any other prior molecular technology. The embracing of a new technology for routine diagnostics usually takes 10 - 14 years or more. Despite its rapid adoption into clinical testing, NGS has a number of intricacies associated with its implementation that are unfamiliar to the clinical laboratory. This talk will address the recently developed CAP/CLIA standards and PT being developed for different parts of the NGS workflow in clinical testing.

11:10 Quantitative Assessment of Tissue Sample Quality
Shannon McCall, M.D., Director, Duke Biospecimen Repository and Processing Core, Pathology, Duke University School of Medicine
CAP Accreditation offers an opportunity for standardization of practice. Today, there are few resources for biobanks that seek external verification/quality assurance of how they are performing and how they are conforming to benchmark standards of quality. The CAP Biorepository Accreditation Program features a unique peer-based inspector model that integrates education and a sharing of best practices to advance quality.

11:40 Improving Clinical Laboratory Quality through Effective Proficiency Testing: Practical Considerations in an Evolving Virtual World
John Osiecki, Ph.D., Director, Scientific Affairs, Roche Molecular Systems
Proficiency testing programs are so important to maintaining acceptable levels of performance in molecular diagnostic laboratories. Addressing current challenges, discussing the newest technologies (sequencing), and what the future holds for quality assurance and proficiency testing are just a few of the exciting topics this presentation will cover.

12:10 pm Session Break

12:20 Luncheon Presentation *(Sponsorship Opportunity Available)* **or Lunch on Your Own**

1:00 Refreshment Break in the Exhibit Hall and Last Chance for Poster Viewing

PRE-ANALYTICAL CONSIDERATIONS IN CANCER GENOMIC ANALYSIS

1:40 Chairperson's Remarks
Jane Emerson, M.D., Ph.D., Professor of Clinical Pathology, University of Southern California

1:45 Biospecimen Quality Appraisal at a Cancer Tissue Bank
Teri A. Longacre, M.D., Professor of Pathology; Director, Tissue, Procurement Facility, Stanford Cancer Center
Quality control of biospecimen banking in a high volume academic cancer center requires a dedicated anatomic pathologist with specific expertise in tumor pathology. Methods to ensure high quality biospecimen banking, storage, and distribution based on best practices and evidence-based standards are presented.

2:15 Quantification of HER2 Heterogeneity in Patient Samples
Elena Geretti, Ph.D., Senior Scientists, Merrimack Pharmaceuticals
The currently FDA-approved methods for HER2 quantification are not able to give a quantitative measure of HER2 heterogeneity of expression. We have developed a novel immunofluorescence assay to quantify HER2 on FFPE samples. Coupled with automated image analysis, our assay is able to quantify HER2 expression at the single cell level, and may constitute a means to understand the predictive and/or prognostic role of the heterogeneity of HER2 expression for patient response to HER2-targeted therapies.

2:45 Sample Type Bias in the Analysis of Cancer Genomes: How Admixed Normal Cells, Intratumoral Heterogeneity, and *ex vivo* Growth Impact Cancer Genomic Analyses
David Solomon, M.D., Ph.D., Anatomic Pathology, PGY-2, University of California, San Francisco
Techniques have emerged which now allow us to interrogate the entire genome of human cancers and to define the genetic alterations that drive tumorigenesis. When performing these genomic analyses, it is important to understand the impact that admixed normal cells, intratumoral heterogeneity, and *ex vivo* growth can have on the results. This talk will discuss these issues and highlight one study comparing genomic analyses performed on glioblastoma tumor samples of differing types (primary tumors, primary xenografts, primary cultures, and established cell lines).

3:15 Q&A with Session Speakers

3:45 Refreshment Break

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BIOSPECIMEN SCIENCE AND SAMPLE PREP

continued

TARGET ENRICHMENT

4:00 Chairperson's Remarks

4:05 Nucleic Acid Target Enrichment from Clinical Samples: Separating the Needle from the Haystack

Michael A. Lewinski, Ph.D., Senior Director, Clinical Research, Microbiology, Roche Molecular Systems, Inc.

Detection of nucleic acid sequences in clinical samples present in low concentration relative to background is important across all disciplines of medicine, including cancer, prenatal diagnosis and infectious diseases. Target enrichment methods concentrating sequences of interest and removing irrelevant nucleic acid and potential inhibitors from clinical samples have been described and have been shown to improve assay sensitivity and patient outcomes. This session focuses on target enrichment methods for improved detection of low copy number targets directly from clinical samples.

BLOOD SPECIMEN ISSUES

4:35 Photopolymer Separators for Standardizing Cell-Fractions in Blood Specimens

Jane Emerson, M.D., Ph.D., Professor of Clinical Pathology, University of Southern California

Photopolymers are incorporated into separator gels in evacuated blood collection tubes in order to create a solid barrier at the interface between serum/plasma and blood cells. We have shown cell-free DNA and mRNA from exosomes is reliably analyzed from these specimens. This technique allows for complete recovery of the cell-free fraction which will enable standardization of low concentration analytes.

5:05 Serum Blood Collection Tube Additive Interference on Clinical Assays

Raffick Bowen, Ph.D., MHA, MT, Clinical Associate Professor of Pathology, Stanford University Medical Center

Components of blood collection tubes, such as stoppers, stopper lubricants, tube walls, surfactants, clot activators, and separator gels may add materials to blood, adsorb components, or interact with protein and cellular components. As a consequence of the multiple and complex interactions of collection devices with blood specimens, collection devices can be a major source of potential error in the pre-analytical phase of laboratory testing. Understanding the interactions of collection devices with blood specimens is essential for the performance of accurate laboratory testing. This presentation will discuss some tube-specific interference on clinical assays, particularly immunoassays.

5:35 Close of Conference Program

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Cambridge Healthtech Institute's Inaugural

CLINICAL EPIGENETICS

Interrogating Disease Epigenomes for Diagnostic, Prognostic & Therapeutic Utility

MONDAY, FEBRUARY 10

10:30 am Conference Program Registration

KEYNOTE SESSION: CANCER (EPI)GENOMICS

11:50 Chairperson's Opening Remarks

12:00 pm The Cancer Epigenome

Peter A. Jones, Ph.D., D.Sc., Director, USC Norris Comprehensive Cancer Center; Distinguished Professor, Urology, Biochemistry & Molecular Biology, Keck School of Medicine, University of Southern California

Recent genome sequencing projects have revealed a surprising number of unanticipated mutations in genes which modify the epigenome. These studies have revealed a much closer interaction between the genetic and epigenetic basis of human cancer. We have begun to explore the potential effects of these mutations on the epigenome, paying particular attention to the existence and roles of chromatin accessibility in carcinogenesis. There is also excitement at the potential to use drugs to target these changes and I will discuss the effects of DNA methylation inhibitors.

12:30 Crosstalk between 5-Methylcytosine, 5-Hydroxymethylcytosine and Histone Modifications in Cancer

Gerd Pfeifer, Ph.D., Lester M. and Irene C. Finkelstein Endowed Chair, Biology & Professor, Epigenetics and Genetics of Cancer, Cancer Biology, City of Hope

Although changes of the epigenome, such as perturbation of DNA methylation patterns, are common events in human cancer, their contribution to the initiation and progression of malignant tumors has remained unclear. We have analyzed histone and DNA cytosine modifications in human cancer and normal tissue. The relationship between chromatin marks and cytosine modifications will be discussed. We propose a model in which specific epigenetic changes are shown to be important for the selection of tumor-driving events and thus may play a major role in cancer progression.

1:00 Session Break

1:15 Luncheon Presentation I: Accelerating Large Scale Genomics and Translational Research Using an Integrated High Performance Computing Solution

Sponsored by IBM

Kathy Tzeng, Ph.D., Team Lead, Life Sciences Solution Enablement, IBM

Scott Markel, Ph.D., Principal Bioinformatics Architect, Accelrys

Advancements in life science research and translational medicine drive the need for new technologies and computing approaches. These approaches pose challenges for IT leaders, researchers and developers to manage, share and store higher data volumes with greater efficiency at lower cost. Life sciences industry experts will discuss compute- and data-intensive challenges, the latest IBM genomic medicine solutions and real-world strategies adopted by leading genomic research institutes for large-scale data projects.

1:45 Luncheon Presentation II Beyond Known microRNAs: Exploring the Rest of the Small RNA Transcriptome

Sponsored by Maverix Biomics

Todd M. Lowe, Ph.D., CSO, Maverix Biomics, Inc.

Within the human transcriptome, microRNAs have proven to be the most dynamic, functionally important class of small non-coding RNAs, influencing the regulation of the majority of protein coding genes. However, largely unexplored small RNA transcriptome data sets, both public (ENCODE, TCGA, SRA) and private, have revealed a very wide range of novel small RNA transcripts with potential to be new biomarkers or uncharacterized regulators. To enable hands-on exploration of RNA-seq data sets by a much larger community of biologists, we have developed the Maverix Analytic Platform, integrating tools and data at the command of any researcher. We show examples of these overlooked classes of small RNAs to illustrate the untapped opportunities for discovery.

2:15 Session Break

KEYNOTE SESSION: BIOTECHS FUELING EPIGENETIC DRUG DISCOVERY

2:30 Chairperson's Remarks

2:35 Talk Title to be Announced

Eric Hedrick, M.D., CMO, Epizyme

3:05 Small Molecule Inhibition of BET Protein Bromodomains: Mechanisms of Action and Potential Therapeutic Applications

Michael R. Cooper, M.D., CMO, Constellation Pharmaceuticals

Constellation and others are now developing small molecule inhibitors of BET protein bromodomains that have pharmaceutical properties suitable for clinical application. Given that the anti-tumor activity of BET inhibitors may arise from the suppression of a number of different cancer-relevant genes, the challenge will be to identify the most important determinants of anti-tumor activity in different malignancies. The presentation will review mechanistic studies of BET inhibitors and discuss potential molecularly defined disease subsets for clinical evaluation.

3:35 Early Clinical Development of OTX015, a Potent BET Bromodomain Inhibitor

Esteban Cvitkovic, M.D., Co-Founder and CSO, OncoEthix

I will present key information on the molecule and clinical formulations of OTX015, providing results from randomized, placebo-controlled trials in healthy volunteers. Also, preliminary results from ongoing dose-range finding studies in patients with relapsed/refractory hematologic malignancies will be discussed.

4:05 Chromatin Remodeling - A Novel Strategy to Control Harmful Alcohol Drinking

Dorit Ron, Ph.D., Professor, Neurology and Endowed Chair, Cell Biology, University of California, San Francisco

4:35 Refreshment Break and Transition to Plenary Keynote

5:00 PLENARY KEYNOTE SESSION
(please see page 4 for details)

6:15 Grand Opening Reception in the Exhibit Hall with Poster Viewing

7:45 Close of Day

TUESDAY, FEBRUARY 11

7:00 am Registration and Morning Coffee

8:00 PLENARY KEYNOTE SESSION
(please see page 4 for details)

9:15 Refreshment Break in the Exhibit Hall with Poster Viewing

Sponsored by [illegible]

NEXT-GENERATION EPIGENETIC TARGETS

CLINICAL EPIGENETICS
CONTINUED ON NEXT PAGE

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CLINICAL EPIGENETICS

continued

10:25 Chairperson's Remarks
Dominique Verhelle, Ph.D., MBA, Director, Epigenetics, Tumor Cell Biology, Oncology Research Unit, Pfizer, Inc.

10:30 Disorders of Histone Methylation in Hematological Malignancy
Jonathan D. Licht, M.D., Johanna Dobe Professor & Chief, Division of Hematology/Oncology, Feinberg School of Medicine, Northwestern University

I will discuss the recurrent mutations in epigenetic enemies affecting the pivotal change in hit one methylation between histone 3 lysine 4 (H3K4me- activation) and H3K27me (repression). Among the proteins that will be discussed are EZH2, which is mutated in a substantial fraction of germinal center derived lymphomas; UTX, a histone demethylase for H3K27 that is deleted in multiple myeloma and other tumors; and WHSC1/MMSET, a gene over expressed in multiple myeloma as well as many solid tumors and mutated in a subset of acute lymphocytic leukemia.

11:00 Critical Roles of Histone Methyltransferases and Demethylases in Human Carcinogenesis
Ryuji Hamamoto, Ph.D., Associate Professor, Hematology & Oncology, The University of Chicago

It has become clear that methyl groups stand beside phosphate groups as major controlling elements in protein function. In this context, we have already identified a number of histone methyltransferases and demethylases, which are related to human cancer. According to our detailed functional analysis, these enzymes relevant to histone methylation are likely to play a crucial role in human carcinogenesis. Development of anti-cancer drugs targeting histone methyltransferases and demethylases appears to be an important approach in cancer treatment.

11:30 SWI/SNF ATP-Dependent Chromatin Remodeling: Novel Avenues for Therapeutic Targeting
Mariela Jaskelioff, Ph.D., Investigator, Oncology, Novartis Institutes for BioMedical Research, Inc.

Epigenetic alterations play a key role in cancer, and epigenetic pathways present new avenues for therapeutic targeting in cancer. Through large scale functional studies targeting the epigenome, we uncovered a robust synthetic lethal relationship involving members of the SWI/SNF chromatin remodeling complex. Cancer cells suffering loss of BRG1, an ATP-dependent subunit of the SWI/SNF complex, are exquisitely dependent on the closely related but distinct SWI/SNF catalytic subunit BRM. BRM is therefore an attractive therapeutic target in BRG1-mutant cancers.

12:00 pm Who Will Benefit from Epigenetic Drugs?
Dominique Verhelle, Ph.D., MBA, Director, Epigenetics, Tumor Cell Biology, Oncology Research Unit, Pfizer, Inc.

One of the problems the industry involved in epigenetic drug discovery currently faces is to identify and to expand the use of the specific inhibitors. Since tumor response to an epigenetic inhibitor may be independent of target expression, mutational status and substrate levels, different approaches may be required to identify responder patients. This presentation will focus on precision medicine approaches applied to epigenetic targets.

12:30 Session Break

12:40 Luncheon Presentation: Creating Predictive Disease Models Using Knowledge Networks
Sponsored by ELSEVIER
Nikolai Daraselia, Ph.D., Director, Research, Elsevier

Drug induced cholestasis is a common liver toxicity resulting in reduced bile acid secretion – a potential adverse event in drug development. A cholestasis disease model was developed predicting FGF 19/15 involvement in bile acid biosynthesis and regulation by FXR and PXR. When FXR and PXR are up-regulated, bile acid biosynthesis is down-regulated resulting in drug-induced cholestasis. Since this publication, FXR, PXR and FGF19/15 roles in drug-induced cholestasis have been clearly established. Discussion on the role of predictive molecular disease models in early discovery will be presented.

1:40 Refreshment Break in the Exhibit Hall with Poster Viewing

TARGETING NON-CODING RNA

2:15 Chairperson's Remarks
Leonard Lipovich, Ph.D., Associate Professor, Center for Molecular Medicine and Genetics, Wayne State University School of Medicine

2:20 microRNA Dysregulation in Cancer
Carlo M. Croce, M.D., Professor and Chair, Department of Molecular Virology, Immunology and Medical Genetics, Human Cancer Genetic Program, The Ohio State University

2:50 Long Non-Coding RNA Genes Shift Human Breast Cancer Cells along the Apoptosis-Proliferation Axis
Leonard Lipovich, Ph.D., Associate Professor, Center for Molecular Medicine and Genetics, Wayne State University School of Medicine

The ENCODE Consortium highlighted the extraordinary abundance of long non-coding RNA (lncRNA) genes in the human genome. We interrogated the estrogen-responsive lncRNA transcriptome of human estrogen receptor alpha positive breast cancer, pinpointing a set of estrogen-induced lncRNAs. Knockdown and overexpression of these lncRNAs, followed by five phenotypic assays indicated they have profound and reproducible phenotypic impacts on human breast cancer cell morphology and growth. These functional lncRNAs should be rationally targeted in cancer therapeutics.

3:20 The Impact of Rare Non-Coding Variants of Gene Expression and Disease
Stephen B. Montgomery, Ph.D., Assistant Professor, Pathology, Genetics & Computer Science, Stanford University School of Medicine

Recent and rapid human population expansion has led to an excess of rare genetic variants that are expected to contribute to an individual's genetic burden of disease risk. Large-scale sequencing studies have highlighted an abundance of rare, deleterious variants within protein-coding sequences. However, in addition to rare protein-coding variants, rare non-coding variants are likely enriched for functional consequences. I will highlight investigations of individual, family and population transcriptomes to identify the extent and impact of rare non-coding variants.

3:50 Sponsored Presentations *(Opportunities Available)*

4:20 Valentine's Day Celebration in the Exhibit Hall with Poster Viewing

5:20 Breakout Discussions in the Exhibit Hall *(see website for details)*

6:30 Close of Day

WEDNESDAY, FEBRUARY 12

7:00 am Breakfast Presentation *(Sponsorship Opportunity Available)* **or Morning Coffee**

» **8:00 PLENARY KEYNOTE SESSION**
(please see page 4 for details)

9:45 Refreshment Break and Poster Competition Winner Announced in the Exhibit Hall

EPIGENETIC BIOMARKERS AND DIAGNOSTICS

10:35 Chairperson's Remarks
Michelle M. Hanna, Ph.D., CEO & Scientific Director, RiboMed Biotechnologies, Inc.

10:40 Identification of Genome-Wide Methylated CpG Island Profiles of Coding, Non-Coding and Repeated Regions as Molecular Markers for

CLINICAL EPIGENETICS
CONTINUED ON NEXT PAGE

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CLINICAL EPIGENETICS

continued

Human Melanomas
Ranjan J. Perera, Ph.D., Associate Professor & Scientific Director, Genomics and Bioinformatics, Sanford-Burnham Medical Research Institute
We have identified genome-wide methylated CpG island distributions by subjecting melanoma genomic DNA to methyl binding domain 2 (MBD2) pull-down and NGS. CpG islands in the upstream regulatory regions of many coding and non-coding RNA genes exhibit extensive hypermethylation, whereas several repeated elements, such as LINE 2, and several LTR elements, are hypomethylated in advanced stage melanoma cell lines. Focused assays of melanoma patient tissue samples for CpG island methylation near the non-coding RNA genes demonstrated high specificity.

11:10 miRNA Biomarkers for Colorectal Neoplasia
Ajay Goel, Ph.D., Director, Epigenetics and Cancer Prevention, Baylor Research Institute
MicroRNAs (or miRNAs) are small transcripts of 20-24 nucleotides that have emerged as important regulators of gene expression in cancer cells. Overexpression of specific miRNAs has been linked to the stepwise disease progression during the normal-adenoma-cancer sequence in the colorectal cancer (CRC). Given their cancer-specific pattern of expression, remarkable stability and presence in blood and other body fluids, miRNAs are considered to be highly promising cancer biomarkers.

11:40 PANEL DISCUSSION: Developing and Commercializing Epigenetic Diagnostics
Moderator: Perry Dimas, VP, Business Development, Premier Source Diagnostics
Panelists:
Michelle M. Hanna, Ph.D., CEO & Scientific Director, RiboMed Biotechnologies, Inc.
Noel Doheny, CEO, Epigenomics, Inc.
Babak Alizadeh, Ph.D., Co-founder & COO, PrognosDx Health, Inc.
Additional Panelists to be Announced
Epigenetic marks, such as DNA methylation and histone modifications, comprise part of the epigenetic machinery leading to abnormal gene expression and chromatin instability in disease. Epigenetic changes, particularly in human cancers, are now being considered as novel biological markers for diagnostic and therapeutic utility. This panel will discuss current challenges in developing and commercializing epigenetic diagnostics by addressing three specific areas of concern:

- DNA Methylation as Viable Biomarkers
- Validation of Assay and Technologies
- CPT Coding and Reimbursement Challenges

12:10 pm Session Break

12:20 Luncheon Presentation *(Sponsorship Opportunity Available)* **or Lunch on Your Own**

1:00 Refreshment Break in the Exhibit Hall and Last Chance for Poster Viewing

EPIGENETIC THERAPEUTICS: PRECLINICAL DEVELOPMENT

1:40 Chairperson's Remarks
Peter Staller, Ph.D., Director, Oncology Research, EpiTherapeutics ApS

1:45 Development of Specific and Reversible Inhibitors of Lysine Specific Demethylase 1 (LSD1)
Sunil Sharma, M.D., Division Chief, Medical Oncology, Jon and Karen Huntsman Professor, Cancer Research, Huntsman Cancer Institute, University of Utah; CMO, Salarius Pharmaceuticals
I will describe the efforts to develop inhibitors of LSD1. LSD1 is an important histone demethylase that is important for histone K4 and K9 demethylation. It has an emerging role in various cancers. In this presentation, I will review the biology and rationale for LSD1 inhibition and various strategies to inhibit its activity.

2:15 Inhibition of LSD1 as a Therapeutic Strategy for the Treatment of Acute Myeloid Leukemia
Helai Mohammad, Ph.D., Investigator, Cancer Epigenetics DPU, Oncology R&D, GlaxoSmithKline
Lysine specific demethylase 1 (LSD1) is a H3K4me1/2 demethylase found in various transcriptional co-repressor complexes. The current study describes the anti-tumor effects of a novel, potent, selective, irreversible GSK LSD1 inhibitor. Our pre-clinical data demonstrate that pharmacological inhibition of LSD1 may provide a promising treatment for AML by promoting differentiation and subsequent growth inhibition of AML cells.

2:45 Co-Presentation: The Constellation-Genentech Bromodomain Drug Discovery Platform
Andrea Cochran, Ph.D., Senior Scientist, Early Discovery Biochemistry, Genentech, Inc.
Steve Bellon, Ph.D., Director, Structural Biology, Constellation Pharmaceuticals
We will describe a platform for discovery of potent and selective bromodomain probes for use in target validation studies. The platform consists of medicinal chemistry, biophysics and structural biology applied to understanding how bromodomains recognize both chromatin and small molecules. Cell-permeable small-molecule inhibitors are useful tools to investigate the function of bromodomain proteins in activating transcription and may provide starting points for the development of novel drugs.

3:15 Mechanisms of Response to BET Bromodomain Inhibition in Malignant Peripheral Nerve Sheath Tumors
Lu Q. Le, M.D., Ph.D., Assistant Professor, Department of Dermatology, Simmons Comprehensive Cancer Center, University of Texas Southwestern Medical Center
Malignant Peripheral Nerve Sheath Tumors (MPNSTs) are highly aggressive sarcomas that are typically fatal. To gain insights into MPNST pathogenesis, we took advantage of a novel MPNST mouse model that permits the study of tumor evolution, which allowed us to identify and elucidate mechanisms for chromatin regulator BRD4 in promoting MPNST tumorigenesis. These findings provide a strong preclinical basis for evaluating BRD4 inhibitors as novel therapies for these life-threatening tumors.

3:45 Refreshment Break

4:05 The Potential Applications of Enzymatic Inhibitors of KDM5 in Oncology
Peter Staller, Ph.D., Director, Oncology Research, EpiTherapeutics ApS
The histone demethylases KDM5A and KDM5B target the methylation of histone H3 at lysine 4 and contribute to cancer cell proliferation and to the induction of drug tolerance. EpiTherapeutics has developed specific and potent inhibitors of KDM5. The pharmacological properties of selected compounds and their *in vivo* activity as well as potential therapeutic applications will be discussed.

EPIGENETIC THERAPEUTICS: CLINICAL DEVELOPMENT

4:35 Resminostat, A Novel Histone Deacetylase Inhibitor in Development for the Treatment of Advanced Hepatocellular Carcinoma (HCC): A New Biomarker-Driven Epigenetic Therapy Option
Bernd Hentsch, Ph.D., Chief Development Officer, 4SC AG
Resminostat has been clinically studied in patients with hepatocellular carcinoma (HCC), Hodgkin's Lymphoma (HL) and colorectal cancer. A biomarker program was established identifying transcriptional profiles modulated upon resminostat exposure. ZFP64 was identified as a biomarker strongly downregulated by resminostat. Expression levels of ZFP64 measured prior to resminostat treatment in peripheral blood cells correlated with the clinical outcome, i.e., patients displaying high ZFP64 baseline levels experienced a substantially longer overall survival.

5:05 Bromodomain Inhibition for Treating Human Diseases
Norman C.W. Wong, M.D., CSO & Co-Founder, Resverlogix
The knowledge gained from 5 years of human clinical trials using RVX-208, an orally active BET-protein inhibitor, has provided valuable information in the design of a discovery platform. New compounds identified using this platform have proven to be active in cellular and animal models of cancerous and inflammatory diseases. The latest data detailing characteristics of novel BET-protein inhibitors will be presented.

5:35 Close of Conference Program

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Cambridge Healthtech Institute's Fifth Annual

GENOME AND TRANSCRIPTOME ANALYSIS

Next-Generation Sequencing of Disease Genomes, Epigenomes & Transcriptomes

MONDAY, FEBRUARY 10

10:30 am Conference Program Registration

» KEYNOTE SESSION: CANCER (EPI)GENOMICS

11:50 Chairperson's Opening Remarks

12:00 pm The Cancer Epigenome

Peter A. Jones, Ph.D., D.Sc., Director, USC Norris Comprehensive Cancer Center; Distinguished Professor, Urology, Biochemistry & Molecular Biology, Keck School of Medicine, University of Southern California

Recent genome sequencing projects have revealed a surprising number of unanticipated mutations in genes which modify the epigenome. These studies have revealed a much closer interaction between the genetic and epigenetic basis of human cancer. We have begun to explore the potential effects of these mutations on the epigenome, paying particular attention to the existence and roles of chromatin accessibility in carcinogenesis. There is also excitement at the potential to use drugs to target these changes and I will discuss the effects of DNA methylation inhibitors.

12:30 Crosstalk between 5-Methylcytosine, 5-Hydroxymethylcytosine and Histone Modifications in Cancer

Gerd Pfeifer, Ph.D., Lester M. and Irene C. Finkelstein Endowed Chair, Biology & Professor, Epigenetics and Genetics of Cancer, Cancer Biology, City of Hope

Although changes of the epigenome, such as perturbation of DNA methylation patterns, are common events in human cancer, their contribution to the initiation and progression of malignant tumors has remained unclear. We have analyzed histone and DNA cytosine modifications in human cancer and normal tissue. The relationship between chromatin marks and cytosine modifications will be discussed. We propose a model in which specific epigenetic changes are shown to be important for the selection of tumor-driving events and thus may play a major role in cancer progression.

1:00 Session Break

Sponsored by IBM

1:15 Luncheon Presentation I: Accelerating Large Scale Genomics and Translational Research Using an Integrated High Performance Computing Solution

Kathy Tzeng, Ph.D., Team Lead, Life Sciences Solution Enablement, IBM

Scott Markel, Ph.D., Principal Bioinformatics Architect, Accelrys

Advancements in life science research and translational medicine drive the need for new technologies and computing approaches. These approaches pose challenges for IT leaders, researchers and developers to manage, share and store higher data volumes with greater efficiency at lower cost. Life sciences industry experts will discuss compute- and data-intensive challenges, the latest IBM genomic medicine solutions and real-world strategies adopted by leading genomic research institutes for large-scale data projects.

Sponsored by Maverix Biomics

1:45 Luncheon Presentation II Beyond Known microRNAs: Exploring the Rest of the Small RNA Transcriptome

Todd M. Lowe, Ph.D., CSO, Maverix Biomics, Inc.

Within the human transcriptome, microRNAs have proven to be the most dynamic, functionally important class of small non-coding RNAs, influencing the regulation of the majority of protein coding genes. However, largely unexplored small RNA transcriptome data sets, both public (ENCODE, TCGA, SRA) and private, have revealed a very wide range of novel small RNA transcripts with potential to be new biomarkers or uncharacterized regulators. To enable hands-on exploration of RNA-seq data sets by a much larger community of biologists, we have developed the Maverix Analytic Platform, integrating tools and data at the command of any researcher. We show examples of these overlooked classes of small RNAs to illustrate the untapped opportunities for discovery.

2:15 Session Break

» KEYNOTE SESSION: ADVANCES IN GENOMIC ANAYLSIS

2:30 Chairperson's Remarks

Kathy Tzeng, Ph.D., Team Lead, Life Sciences Solution Enablement, IBM

2:35 What Does Our Genome ENCODE?

John Stamatoyannopoulos, M.D., Associate Professor, Genome Sciences and Medicine, School of Medicine, University of Washington

3:05 Pan-Cancer to Personalized Pathway Targets

Josh Stuart, Ph.D., Baskin Engineering Endowed Chair & Professor, Department of Biomolecular Engineering; Associate Director, Center for Biomolecular Science & Engineering, University of California, Santa Cruz

The Cancer Genome Atlas Research Network has profiled and analyzed large numbers of human tumors to discover molecular aberrations at the DNA, RNA, protein and epigenetic levels. The rich data resulting from that enterprise provides a major opportunity to develop a picture of commonalities, differences, and emergent themes across tumor lineages. In this talk I will discuss our efforts to create new tools to elucidate disrupted pathways in cancer cells to infer the "health" of cellular circuitry.

3:35 Single Cell Genome Sequencing: Life at the Single Molecule Level

Xiaoliang Sunney Xie, Ph.D., Mallinckrodt Professor, Chemistry, Department of Chemistry and Chemical Biology, Harvard University

Point mutation and copy number variation, which are two major dynamical changes of DNA, can now be studied at the single cell level by whole genome amplification and sequencing. Experiments probing the biology of meiosis and cancer will be described. We demonstrate the proof of principle of selecting oocytes in *in vitro* fertilization in order to avoid miscarriage and genetic diseases. We also show that individual circulating tumor cells can be sequenced, providing tumor genetic signatures for personalized therapy.

4:05 Sponsored Presentations *(Opportunities Available)*

4:35 Refreshment Break and Transition to Plenary Keynote

» 5:00 PLENARY KEYNOTE SESSION
(please see page 4 for details)

6:15 Grand Opening Reception in the Exhibit Hall with Poster Viewing

7:45 Close of Day

GENOME AND TRANSCRIPTOME ANALYSIS
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GENOME AND TRANSCRIPTOME ANALYSIS

continued

TUESDAY, FEBRUARY 11

7:00 am Registration and Morning Coffee

8:00 PLENARY KEYNOTE SESSION
(please see page 4 for details)

9:15 Refreshment Break in the Exhibit Hall with Poster Viewing

Sponsored by

SINGLE-CELL GENOME & TRANSCRIPTOME SEQUENCING

10:25 Chairperson's Remarks

Jan Vijg, Ph.D., Professor & Chair, Department of Genetics, Albert Einstein College of Medicine

10:30 Sequencing Both the Transcriptome and Genome of Single Cells in Health and Disease

Chris P. Ponting, Ph.D., Head, Computational and Disease Genomics, Functional Genomics Unit, University of Oxford; Associate Member, Wellcome Trust Sanger Institute

In this presentation I will discuss experimental approaches being developed to sequence the poly-adenylated transcriptome together with the genome of individual cells. These approaches now enable consideration of the effects of DNA variants on transcript levels in single cells.

11:00 Dynamic Heterogeneity of Primary Immune Cells Revealed By Large-Scale Single-Cell RNA-Seq

Hongkun Park, Ph.D., Professor, Chemistry and Physics, Department of Chemistry and Chemical Biology, Harvard University; Associate Member, The Broad Institute

Recent molecular studies have revealed that individual cells can exhibit substantial differences in gene expression, protein levels, and phenotypic output, with important functional consequences. I will describe our efforts to dissect this heterogeneity and its biological implications using large-scale single-cell transcriptomics. In particular, I will discuss our recent studies of immune dendritic cells and T cells using single-cell RNA-Seq and describe how these studies can be used to uncover functional diversity between cells and to decipher cell states and circuits.

11:30 Single-Cell Genomics in Aging

Jan Vijg, Ph.D., Professor & Chair, Department of Genetics, Albert Einstein College of Medicine

To dissect age-related intra-tissue heterogeneity we developed single-cell, genome-wide sequencing procedures to measure both single nucleotide and structural variation. To directly link single-cell, genomic mutation loads to possible consequences at the level of the transcriptome, we performed concurrent global mRNA and whole genome amplification, followed by RNA-Seq and whole exome sequencing. This method, "single-cell transcriptogenomics", allows us to directly track the consequences of randomly induced genetic mutations on gene expression profiles in the same single cell.

12:00 pm Genetic Programming in Human and Mouse Early Embryos Revealed by Single-Cell RNA Sequencing

Guoping Fan, Ph.D., Professor, Human Genetics, University of California, Los Angeles

We report here a comprehensive analysis of transcriptome dynamics from oocyte to morula in both human and mouse embryos, using single-cell RNA sequencing. By weighted gene co-expression network analysis, we find that each developmental stage can be delineated concisely by a small number of functional modules of co-expressed genes. This result indicates a sequential order of transcriptional changes in pathways of cell cycle, gene regulation, translation and metabolism, acting in a step-wise fashion from cleavage to morula.

12:30 Session Break

Sponsored by

12:40 Luncheon Presentation I: Creating Predictive Disease Models Using Knowledge Networks

Nikolai Daraselia, Ph.D., Director, Research, Elsevier

Drug induced cholestasis is a common liver toxicity resulting in reduced bile acid secretion – a potential adverse event in drug development. A cholestasis disease model was developed predicting FGF 19/15 involvement in bile acid biosynthesis and regulation by FXR and PXR. When FXR and PXR are up-regulated, bile acid biosynthesis is down-regulated resulting in drug-induced cholestasis. Since this publication, FXR, PXR and FGF19/15 roles in drug-induced cholestasis have been clearly established. Discussion on the role of predictive molecular disease models in early discovery will be presented.

1:10 Luncheon Presentation II *(Sponsorship Opportunity Available)*

1:40 Refreshment Break in the Exhibit Hall with Poster Viewing

FUNCTIONAL IMPACT OF NON-CODING RNA

2:15 Chairperson's Remarks

Leonard Lipovich, Ph.D., Associate Professor, Center for Molecular Medicine and Genetics, Wayne State University School of Medicine

2:20 microRNA Dysregulation in Cancer

Carlo M. Croce, M.D., Professor and Chair, Department of Molecular Virology, Immunology and Medical Genetics, Human Cancer Genetic Program, The Ohio State University

2:50 Long Non-Coding RNA Genes Shift Human Breast Cancer Cells along the Apoptosis-Proliferation Axis

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The ENCODE Consortium highlighted the extraordinary abundance of long non-coding RNA (lncRNA) genes in the human genome. We interrogated the estrogen-responsive lncRNA transcriptome of human estrogen receptor alpha positive breast cancer, pinpointing a set of estrogen-induced lncRNAs. Knockdown and overexpression of these lncRNAs, followed by five phenotypic assays indicated they have profound and reproducible phenotypic impacts on human breast cancer cell morphology and growth. These functional lncRNAs should be rationally targeted in cancer therapeutics.

3:20 The Impact of Rare Non-Coding Variants of Gene Expression and Disease

Stephen B. Montgomery, Ph.D., Assistant Professor, Pathology, Genetics & Computer Science, Stanford University School of Medicine

Recent and rapid human population expansion has led to an excess of rare genetic variants that are expected to contribute to an individual's genetic burden of disease risk. Large-scale sequencing studies have highlighted an abundance of rare, deleterious variants within protein-coding sequences. However, in addition to rare protein-coding variants, rare non-coding variants are likely enriched for functional consequences. I will highlight investigations of individual, family and population transcriptomes to identify the extent and impact of rare non-coding variants.

3:50 Sponsored Presentations *(Opportunities Available)*

4:20 Valentine's Day Celebration in the Exhibit Hall with Poster Viewing

5:20 Breakout Discussions in the Exhibit Hall *(see website for details)*

6:30 Close of Day

GENOME AND TRANSCRIPTOME ANALYSIS
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GENOME AND TRANSCRIPTOME ANALYSIS

continued

WEDNESDAY, FEBRUARY 12

7:00 am Breakfast Presentation *(Sponsorship Opportunity Available)* **or Morning Coffee**

8:00 PLENARY KEYNOTE SESSION
(please see page 4 for details)

9:45 Refreshment Break and Poster Competition Winner Announced in the Exhibit Hall

CANCER GENOMICS: INTEGRATIVE AND COMPUTATIONAL APPROACHES

10:35 Chairperson's Remarks
Francisco M. De La Vega, D.Sc., Visiting Instructor, Department of Genetics, Stanford University School of Medicine

10:40 Concomitant Clonal Evolution of Leukemia Across the Genome, Epigenome, and Transcriptome
Christopher Mason, Ph.D., Assistant Professor, Computational Biomedicine, Weill Cornell Medical College
We have examined patients with acute myelogeneous leukemia (AML) during treatment with genome sequencing, epigenetic profiling (RRBS), and ribo-depleted RNA sequencing. Our results demonstrate a convergence of disrupted biological networks in DNA, RNA, and DNA methylation that mediate chemo-resistance. Also, we observe new prognostic features of epigenetic clones that can predict relapse time, allele-specific switching of damaging mutations, and completely novel, complex genetic rearrangements. Together, these data reveal an extremely dynamic evolutionary landscape of molecular changes, which tumors rapidly engage to survive.

11:10 From Outlier Cancer Phenotypes to Precision Genotypes
Barry S. Taylor, Ph.D., Assistant Professor, Helen Diller Family Comprehensive Cancer Center, University of California, San Francisco
Little is known about the basis of elusive exceptional responses to cancer therapy. I will describe their outlier phenotype-to-genotype approach that uses both whole-genome and deep targeted sequencing to identify the molecular genetic basis of complete and durable response to both targeted and systemic cancer therapies. Recent work will be discussed on the use of such approaches coupled to nuanced computational analyses to identify not only individual sensitizing mutations, but also synergistically acting genetic interactions and the contribution of tumor clonality to the durability of treatment response.

11:40 Computational Approaches for Analyzing Mutational Heterogeneity in Cancer Genomes
Ben Raphael, Ph.D., Associate Professor, Department of Computer Science & Center for Computational Molecular Biology, Brown University
Recent cancer sequencing studies have demonstrated extensive mutational heterogeneity within some tumors, with multiple subpopulations of tumor cells having different complements of somatic mutations. We describe computational techniques to characterize this intra-tumor heterogeneity from whole-genome sequencing data.

12:10 pm Session Break

12:20 Luncheon Presentation *(Sponsorship Opportunity Available)* **or Lunch on Your Own**

1:00 Refreshment Break in the Exhibit Hall and Last Chance for Poster Viewing

COMPUTATIONAL BIOLOGY: TRENDS IN DATA ANALYSIS, INTEGRATION AND VISUALIZATION

1:40 Chairperson's Remarks

1:45 Assembly and Variant Calling of High-Throughput Sequencing Data: Lessons from Large-Scale Sequencing Projects and Moving to the Clinic
Francisco M. De La Vega, D.Sc., Visiting Instructor, Department of Genetics, Stanford University School of Medicine
Here I discuss the principles to implement and validate such pipelines in terms of sensitivity and specificity of the full spectrum of genetic variation, turn-around time and quality, and what are the current open challenges based on experience gained at the 1000 Genomes Project and other large-scale sequencing studies. I will also discuss advanced methods to improve variant quality by leveraging family & population structures, individual germline references in cancer sequencing, and integrative analysis of orthogonal NGS data sets.

2:15 Novel Computational Methods for RNA-Seq Data Analysis
Colin N. Dewey, Ph.D., Associate Professor, Departments of Biostatistics & Medical Informatics and Computer Sciences, University of Wisconsin, Madison
One revolutionary aspect of RNA-Seq is its ability to provide information regarding transcript abundances in species whose genomes have not yet been sequenced. In such situations, transcript quantification involves a combination of *de novo* assembly and abundance estimation, both of which are challenging tasks when a reference genome is unavailable. I will present my group's recent work in addressing these challenges. In particular, our novel methodology for evaluating the accuracy of *de novo* transcriptome assemblies and performing quantification with them.

2:45 Genomics & Sequencing Data Integration, Analysis and Visualization
Carl Meinhof, Manager, Research Informatics, IT, Ceres, Inc.
Scientists expect to navigate genomic data with the same ease and speed that they can navigate geographic data. We have developed a genome browser that uses algorithms from game development to provide high-performance visualization of genomics data. Data from multiple sources can be integrated in a relational database backend, but users can also visualize data from files. Due to its high speed and ease of use the browser facilitates playful exploration of data.

3:15 Sponsored Presentations *(Opportunities Available)*

3:45 Refreshment Break

CLINICAL-GRADE SEQUENCING AND DATA ANALYSIS

4:00 Chairperson's Remarks
Gabe Rudy, Vice President, Product Development, Golden Helix

4:05 CLIA-Certified Cancer Genome Diagnostic Testing in a Rapidly Changing Environment
David Wheeler, Ph.D., Co-Director, Bioinformatics; Associate Professor, Department of Molecular and Human Genetics, Baylor College of Medicine
A CLIA lab seeks reproducibility and stability in the assays performed. However, the rapid accumulation of novel and exciting cancer genome discoveries is expanding our view of the genetic landscape of cancer and impacting how diagnostic testing can be performed. Here I will discuss analytical and biological advances that are changing the way we think about diagnostic services, and how we are integrating them into the CLIA lab.

4:35 Implementation of Next-Generation Sequencing in the Clinical Molecular Oncology Laboratory
Eric Duncavage, M.D., Assistant Professor; Director, Molecular Genetic Pathology Training Program, Department of Pathology and Immunology, Division of Clinical and Genomic Medicine, Washington University
This talk will focus on the design, informatics, and validation of clinical grade NGS-based oncology testing based on experience in the Washington University Genomics and Pathology Services Laboratory. Design considerations including enrichment methods and selecting the best genes for an oncology panel will be discussed and an evaluation of clinical-grade variant analysis tools will be provided. Finally, NGS validation practices, regulatory guidelines, and reimbursement issues will be reviewed.

5:05 Interpreting My DTC Exomes Using Public Access Clinical Databases
Gabe Rudy, Vice President, Product Development, Golden Helix
23andMe provided through a limited pilot the delivery of uninterpreted exomes in 2012. In this talk, I use GenomeBrowse and publicly available clinical databases such as ClinVar and OMIM, as well as many other integrative genomic annotations, to interpret the exomes of myself, wife and son. Recent improvements in alignment, variant calling and public annotation sources may be able to take my research grade exome to a clinical grade interpretation.

5:35 Close of Conference Program

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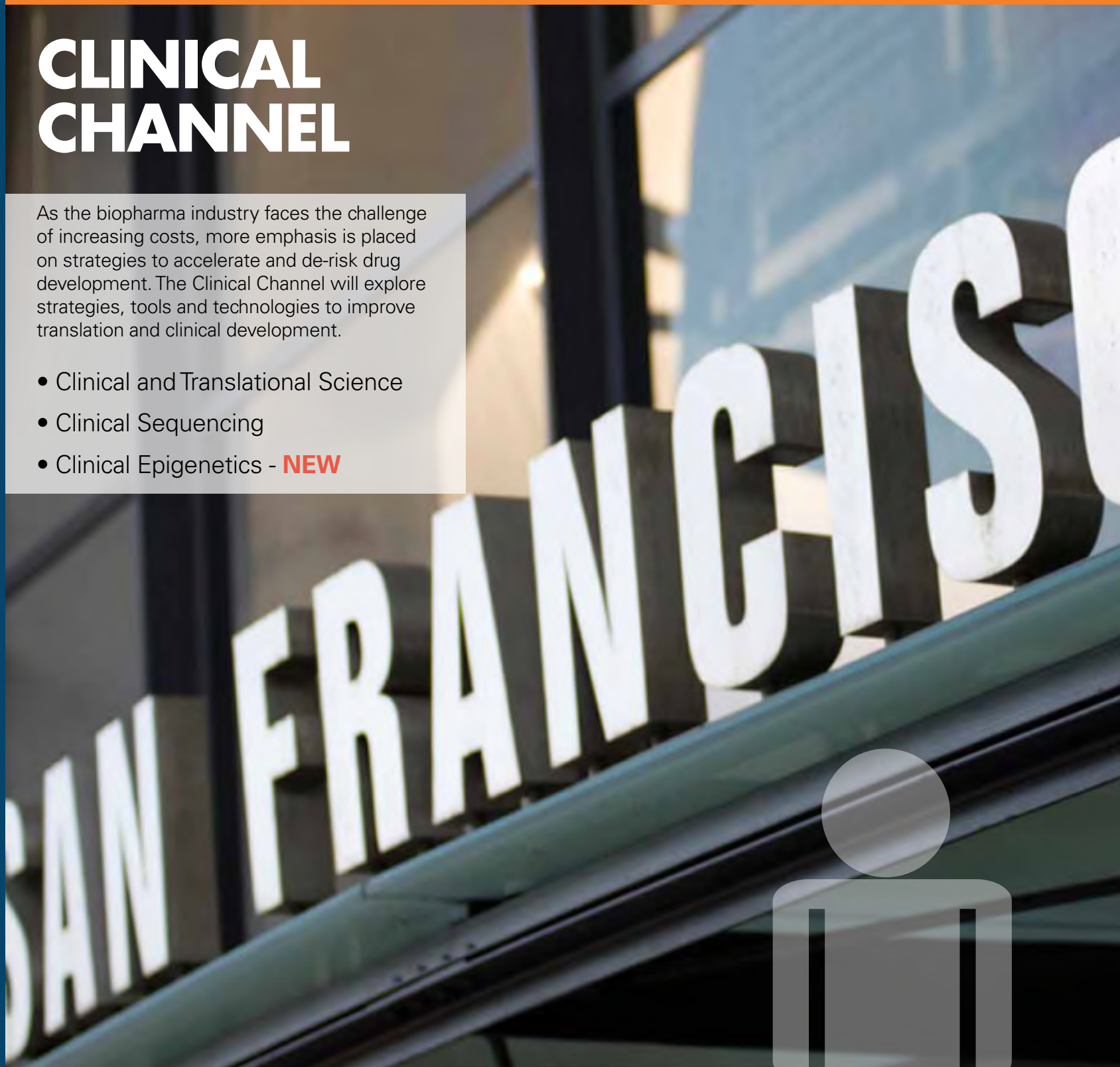
CLINICAL CHANNEL



As the biopharma industry faces the challenge of increasing costs, more emphasis is placed on strategies to accelerate and de-risk drug development. The Clinical Channel will explore strategies, tools and technologies to improve translation and clinical development.

- Clinical and Translational Science
- Clinical Sequencing
- Clinical Epigenetics - **NEW**

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Cambridge Healthtech Institute's Tenth Annual

CLINICAL AND TRANSLATIONAL SCIENCE

Strategies to Accelerate and De-Risk Clinical Development

MONDAY, FEBRUARY 10

10:30 am Conference Program Registration

IMPROVING DECISION MAKING IN DRUG DEVELOPMENT

11:50 Chairperson's Opening Remarks
Lucette Doessegger, M.D., Global Head, Safety Science Licensing & Early Development, F. Hoffmann-La Roche AG

» FEATURED PRESENTATION:

12:00 pm Reinventing Drug Development: Acceleration in the Information Age
Jamie Freedman, M.D., Ph.D., Senior Vice President and Global Head, Clinical Development, MedImmune/AstraZeneca
Time is crucial! The information age provides access to patient-level genomic data; are your development plans keeping pace? Small, targeted studies with strong science are driving development. New thinking, informed patients and strong relationships between academic sites and industry will drive the next generation of blockbusters. Are you in?

12:30 Informing Early Development Decisions
Richard Scheyer, M.D., President and CSO, in vivo veritas, LLC
PoC studies are often used to determine go/no-go. Alternatively, early development may be viewed as the accumulation of knowledge informing probability of successful development, until sufficient to either transition to late phase or to terminate development. For maximal efficiency, early phase should focus on activities having greatest impact on PoS estimation.

1:00 Session Break

1:15 Luncheon Presentation
The Big Catch: Unbiased Trawling for Protein Biomarkers
Sponsored by SomaLogic
Stephen Williams, M.D., Ph.D., CMO, SomaLogic Inc

1:45 Session Break

INNOVATION IN CLINICAL TRIAL DESIGN: MOVING TOWARD PERSONALIZED MEDICINE

2:30 Chairperson's Remarks
Lynn Zieske, Ph.D., Vice President, Commercial Solutions, Life Sciences Business Development, Singulex, Inc.

2:35 Shaping Diagnostic Practice to Drug Development—Moving Beyond Biomarkers
Chris Chamberlain, M.D., Ph.D., Vice President & Head, Experimental Medicine and Diagnostics, UCB Pharma

3:05 Phase II/III Biomarker-Driven Master Protocol for Second Line Therapy of Squamous Cell Lung Cancer (SCCA) *(recorded presentation)*
Vassiliki Papadimitrakopoulou, M.D., Professor, Thoracic/Head & Neck Medical Oncology, University of Texas MD Anderson Cancer Center

3:35 Concept of Translational Safety Medicine and Integrated Safety Management
Lucette Doessegger, M.D., Global Head, Safety Science Licensing & Early Development, F. Hoffmann-La Roche AG

4:05 Multiplex Protein Biomarkers in Clinical Studies: A Strategy for Pharmacodynamic and Companion Diagnostic Discovery
Sponsored by MYRIAD RBM
Dominic Eisinger, Ph.D., Director, Strategic Development, Myriad RBM
The "RBM Approach" to biomarker discovery and validation is one of the most successful means of clinical biomarker analysis for multiple therapeutic areas. Myriad RBM's multiplexed immunoassays are validated to clinical laboratory standards and can quantify hundreds of biomarkers.

4:20 Quantitative Measurement of Low-Abundance Biomarkers Using Digital Single Molecule Counting
Sponsored by Singulex
Lynn R. Zieske, Ph.D., Vice President, Commercial Solutions, Life Sciences Business Development, Singulex, Inc.
Several considerations must be taken into account to establish a clinical biomarker, including the ability to measure normal states. Singulex's proprietary single molecule counting technology helps overcome challenges in biomarker translation from discovery to clinic. This presentation reviews cardiac troponin I (cTnI) development as a clinically relevant biomarker.

4:35 Refreshment Break and Transition to Plenary Keynote

» 5:00 PLENARY KEYNOTE SESSION
(please see page 4 for details)

6:15 Grand Opening Reception in the Exhibit Hall with Poster Viewing

7:45 Close of Day

TUESDAY, FEBRUARY 11

7:00 am Registration and Morning Coffee

» 8:00 PLENARY KEYNOTE SESSION
(please see page 4 for details)

9:15 Refreshment Break in the Exhibit Hall with Poster Viewing
Sponsored by

REDUCING LATE-STAGE ATTRITION THROUGH BETTER SAFETY, EFFICACY, AND POC STUDIES

10:25 Chairperson's Remarks
Manish Muzumdar, Senior Vice President, Products, Remedy Informatics

10:30 Translational Science Approaches to Reduce Phase III Attrition
Kalpana Merchant, Ph.D., CSO, Tailored Therapeutics, Neuroscience, Eli Lilly

11:00 Translational Safety of Immunostimulatory and ADC Cancer Biologics
Rakesh Dixit, Ph.D., DABT, Vice President, R&D; Global Head, Biologics Safety Assessment, MedImmune
With technological advances in generation of immune-stimulatory and armed biologics, the translational safety and toxicology predictions face unprecedented challenges. There is now renewed focus on a better understanding of translational immune-pharmacology associated toxicities (both on-target and off-target). Case studies with major considerations such as target distribution, target pharmacology, and systemic pharmacokinetics, early screening, both traditional and non-traditional (e.g., transgenic models), and *in vitro* safety assessments will be addressed.

11:30 Talk Title to be Announced
Kirk Bertelsen, Ph.D., Director, Clinical Pharmacology, Janssen Pharmaceutical R&D

12:00 pm Translational Value of Target-Based Risk Assessment
Alexander Fekete, Scientific Technical Leader, Novartis

CLINICAL AND TRANSLATIONAL SCIENCE
CONTINUED ON NEXT PAGE

CLINICAL AND TRANSLATIONAL SCIENCE

continued

Early safety risk assessment integrates data from pharmacology and ADME profiling, with later addition of pharmacokinetic experiments and efficacy models. One important element of this approach is target-based *in vitro* profiling for mapping off-target effects to clinical adverse reactions. Ultimately the goal is to produce safer medications for patients and to reduce late-stage attrition through effective translation of predicted toxicological effects.

12:30 Session Break

12:40 Luncheon Presentations *(Sponsorship Opportunities Available)* **or Lunch on Your Own**

1:40 Refreshment Break in the Exhibit Hall with Poster Viewing

SPECIMEN CONSIDERATIONS IN BIOMARKER-DRIVEN CLINICAL TRIALS

2:15 Chairperson's Remarks
Michael H.A. Roehrl, M.D., Ph.D., Director, UHN Program in BioSpecimen Sciences, University of Toronto

2:20 Use of Clinical Genetic Specimens to Support Clinical Drug Development
Rebecca Blanchard, Ph.D., Executive Director, Clinical Research; Head, Clinical Genetics, Merck & Co., Inc.
Many pharmaceutical companies maintain biorepositories of clinical specimens consented for and collected during clinical trials. This presentation will address common uses of biorepository samples to support clinical drug development, and will highlight challenges in specimen collection that impact ultimate utility of the specimen.

2:50 Biospecimen Sample Integrity and Validation of Biomarkers in Clinical Trials
Lokesh Agrawal, Ph.D., Program Director, Biorepositories and Biospecimen Research Branch (BBRB), Cancer Diagnosis Program, Division of Cancer Treatment and Diagnosis, National Cancer Institute

3:20 Best Practices for Management of Clinical Specimen to Support Clinical Drug Development
George Y. Tokiwa, Ph.D., Associate Principal Scientist, Clinical Genetics, Merck Sharp & Dohme Corp.
Many pharmaceutical companies maintain biorepositories of clinical specimens collected during clinical studies that are consented for future biomedical research. This presentation will focus on best practices to address the end to end clinical specimen management activities required to support optimal and compliant use of the specimens.

3:50 When Worlds Collide: Sharing A Vision for an Integrated Clinical Trial Management System *Sponsored by* REMEDY INFORMATICS
Bruce Pharr, Vice President, Product Marketing Laboratory Systems, Remedy Informatics
Mr. Pharr will discuss data strategies to facilitate a more efficient flow of information enterprise-wide between disparate, yet key players in life sciences research and healthcare—such as clinical trial teams, hospitals, labs, biobanks, and others—in order to deliver on and advance the promise of personalized medicine.

4:20 Valentine's Day Celebration in the Exhibit Hall with Poster Viewing

5:20 Breakout Discussions in the Exhibit Hall *(see website for details)*

6:30 Close of Day

WEDNESDAY, FEBRUARY 12

7:00 am Breakfast Presentation *(Sponsorship Opportunity Available)* **or Morning Coffee**

» **8:00 PLENARY KEYNOTE SESSION** *(please see page 4 for details)*

9:45 Refreshment Break and Poster Competition Winner Announced in the Exhibit Hall

CLINICAL AND TRANSLATIONAL BIOMARKERS

10:35 Chairperson's Remarks
Chris Chamberlain, M.D., Ph.D., Vice President & Head, Experimental Medicine and Diagnostics, UCB Pharma

10:40 Making Use of "Big Data": Identification and Modeling of Critical States in Disease Progression on the Example of Chronic Myeloid Leukemia
Andreas Schuppert, Ph.D., Vice President, Technology Development, Bayer Technology Services GmbH; Professor, AICES, RWTH Aachen University

11:10 Translational Science and Predictive Biomarkers for Multiple Receptor Tyrosine Kinase Inhibitor
Yasuhiro Funahashi, Ph.D., Senior Director, Biomarkers and Personalized Medicine, Eisai

11:40 Translating Biomarkers into Companion Diagnostics
Philip Brohawn, Manager, Research & Development, Translational Science, MedImmune

12:10 pm Session Break

12:20 Luncheon Presentation *(Sponsorship Opportunity Available)* **or Lunch on Your Own**

1:00 Refreshment Break in the Exhibit Hall and Last Chance for Poster Viewing

1:40 Chairperson's Remarks

1:45 Late Breaking Presentation

2:15 NOTCH Gene Rearrangement as a Biomarker for Response to GSI Therapy in Breast Cancer
Serguei Lejnine, Ph.D., Associate Director, Merck Research Labs
NOTCH gene translocation resulted in copy number variation between the exons encoding the N-terminal and C-terminal regions of NOTCH. NOTCH translocations were prevalent in the triple negative sub-type (TNBC) of breast cancer (6/66). TNBC cell lines and xenografts with NOTCH translocations are remarkably sensitive to treatment with the gamma-secretase inhibitor (GSI) MRK-003.

CLINICAL AND TRANSLATIONAL SCIENCE
CONTINUED ON NEXT PAGE

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CLINICAL AND TRANSLATIONAL SCIENCE

continued

2:45 Panel Discussion: Biomarker Utility in Accelerating and De-Risking Drug Development

Panelists:

Chris Chamberlain, M.D., Ph.D., Vice President and Head, Experimental Medicine and Diagnostics, UCB Pharma

Philip Brohawn, Manager, Research & Development, Translational Science, MedImmune

Andreas Schuppert, Ph.D., Vice President, Technology Development, Bayer Technology Services GmbH; Professor, AICES, RWTH Aachen University

Suso Platero, Ph.D., Director, Oncology Biomarkers, Janssen Pharmaceuticals

3:45 Refreshment Break

FROM BIG DATA TO TRANSLATIONAL INFORMATICS

4:00 Chairperson's Remarks

Shoibal Datta, Ph.D., Director, Data Sciences, Biogen Idec

4:05 Designing and Building a Data Sciences Capability to Support R&D and Corporate Big Data Needs

Shoibal Datta, Associate Director, R&D Information Technology, Biogen Idec

To achieve Biogen Idec's strategic goals, we have built a cross-disciplinary team to focus on key areas of interest and the required capabilities. To provide a reusable set of IT services we have broken down our platform to focus on the Ingestion, Digestion, Extraction and Analysis of data. In this presentation, we will outline how we brought focus and prioritization to our data sciences needs, our data sciences architecture, lessons learned and our future direction.

4:35 Translational Informatics: Decomposing to Singularity

John Shon, M.D., Head, Translational Informatics IT

There has been an explosion of data across discovery, development, and beyond and all informatics groups are struggling with major challenges in computation, storage and analysis. In a large pharmaceutical environment, the value propositions of informatics lie primarily in three dimensions which I describe. In the larger hyperdynamic environments of research technologies, information technologies, and modern science, interdisciplinary and collaborative approaches become imperative to execute translational strategies effectively.

5:05 Integrating Translational Research Tools

Erik Bierwagen, Ph.D., Principal Programmer Analyst, Department of Bioinformatics, Genentech, Inc.

This talk will cover our efforts at creating an integrated informatics system for animal studies from birth to death and beyond. Our efforts span many different disciplines and groups, but share the common effort of integrating data seamlessly.

5:35 Close of Conference Program

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Cambridge Healthtech Institute's Second Annual

CLINICAL SEQUENCING

Translating NGS to Practice

MONDAY, FEBRUARY 10

10:30 am Conference Program Registration

11:50 Chairperson's Opening Remarks
Julien N. Bradley, MBA, Senior Director, Sales & Marketing, Quanterix Corporation

» 12:00 pm KEYNOTE PANEL DISCUSSION:

CPT Coding

Co-Organized with AMP Association for Molecular Pathology

Moderator: Jill Hagenkord, M.D., CMO & Senior Vice President, InVitae (2013 AMP Economic Affairs Committee and 2014 AMP Training and Education Committee)

Elaine Lyon, Ph.D., Medical Director, Molecular Genetics, ARUP (2014 AMP President and 2013 AMP Economic Affairs, Professional Relations, Strategic Opportunities Committees)

Chris L. Jagmin, M.D., Senior Medical Director, National Medical Policy and Operations, Aetna

- Overview of CPT codes
- Transitioning to the molecular pathology codes from the clinical laboratory's perspective
- Understanding rationale behind new CPT codes

1:00 Session Break

1:15 Luncheon Presentation I: Simoa HD-1: A Fully Automated, Multiplexed Immunoanalyzer with Single Molecule Sensitivity

Sponsored by Quanterix

David C. Duffy, Ph.D., CTO, Quanterix Corporation

Single Molecule Array (Simoa) technology allows multiple proteins to be detected at concentrations 1000-fold lower than currently possible. Simoa is based on the capture of single molecules on paramagnetic beads, and their detection in arrays of femtoliter wells. We will illustrate the power of this analytical sensitivity in diagnosing cancer, neurological diseases, and infectious diseases using the Simoa HD-1 Analyzer, a fully automated instrument designed for use in clinical research and diagnosis.

1:45 Luncheon Presentation II A Novel Exosome RNA Extraction Platform Enabling Biomarker Discovery and Diagnostic Development for Personalized Medicine

Sponsored by exosome diagnostics

Johan Skog, Ph.D., CSO, Exosome Diagnostics

Exosomes are released by cells as an active process of communication, and contain stable, intact nucleic acids, making them an ideal source for biomarker discovery and diagnostic development. ExoRNeasy was optimized to extract RNA, including mRNA, microRNAs, and other RNAs, from plasma and serum. The high quality RNA generated from the ExoRNeasy kit enables profiling of tumor associated mutations as well as RNA levels in biofluids of cancer patients.

2:15 Session Break

PAST, PRESENT AND FUTURE OF PRENATAL GENOMIC TESTING

2:30 Chairperson's Remarks
Daniel H. Farkas, Ph.D., HCLD, FACB, Laboratory Director, Sequenom Center for Molecular Medicine

2:35 Noninvasive Prenatal Testing by Maternal Plasma DNA Sequencing: From Aneuploidy Testing to Fetal Whole Genome and Methylome Sequencing
Dennis Lo, M.D., Ph.D., Director, Li Ka Shing Institute of Health Sciences, Chinese University of Hong Kong

Over the last 5 years, there is much interest in the use of massively parallel sequencing of plasma DNA for noninvasive prenatal testing. Fetal chromosomal aneuploidies, genome-wide molecular karyotyping and even fetal whole genome sequencing were accomplished through the sequencing of maternal plasma DNA. We have shown that this approach can be used for noninvasive determination of the fetal methylome. This latter development has opened exciting opportunities for prenatal testing and research.

3:05 Noninvasive Fetal RHD Genotyping
Daniel H. Farkas, Ph.D., HCLD, FACB, Laboratory Director, Sequenom Center for Molecular Medicine

Antepartum anti-D immunoprophylaxis is standard of care in pregnancy management but is administered unnecessarily to the approximately 40% of mothers who subsequently deliver Rh-negative babies. Clinical laboratory investigation using circulating, cell-free fetal DNA as an analyte provides the potential for more rational management of Rh-negative pregnant women.

3:35 Noninvasive Prenatal Testing 2014: The Basics and Beyond
Christopher Robinson, M.D., MSCR, Associate Professor, Maternal Fetal Medicine, Obstetrics and Gynecology, University of Virginia

This presentation will present the current, state-of-the-art in applied screening and diagnostics involving cffDNA. A focus on the understanding of integration of bench science and bioinformatics will provide the attendee with an expanded understanding of the utilization of cffDNA as a substrate for clinical information and decision-making.

4:05 Clinician-Friendly Tools and Efficient Database Architecture to Accelerate Genetic Diagnoses of Challenging Patients

Sponsored by NEXTCODE HEALTH

Jeff Gulcher, M.D., Ph.D., President, CSO, NextCODE Health

NextCODE offers informatics systems originally developed at deCODE Geneticsalong with a curated knowledge base for end-to-end analysis of whole exome and whole genome data for patients, families, and large cohorts. Clinical Sequence Analyzer (CSA) is a clinician-friendly web-based interface for sequence-based diagnostics. NextCODE's GOR database infrastructure allows for more efficient storage, query, and display of large scale sequence and coverage data. Our *de novo* mutation detector improves accuracy of *de novo* mutation calls.

4:35 Refreshment Break and Transition to Plenary Keynote

» 5:00 PLENARY KEYNOTE SESSION
(please see page 4 for details)

6:15 Grand Opening Reception in the Exhibit Hall with Poster Viewing

7:45 Close of Day

TUESDAY, FEBRUARY 11

7:00 am Registration and Morning Coffee

» 8:00 PLENARY KEYNOTE SESSION
(please see page 4 for details)

9:15 Refreshment Break in the Exhibit Hall with Poster Viewing

Sponsored by

SAMPLE PREP CONSIDERATIONS FOR NGS-BASED CLINICAL ASSAYS

10:25 Chairperson's Remarks
Martin Siaw, Ph.D., Associate Scientific Director, Advanced Sequencing, Quest Diagnostics Nichols Institute

CLINICAL SEQUENCING
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CLINICAL SEQUENCING

continued

10:30 NGS-Based Clinical Assays: Building Castles in the Air

Jamie L. Platt, Ph.D., Scientific Director, Advanced Sequencing, Quest Diagnostics Nichols Institute

While applying NGS in the clinical setting may seem like "building castles in the air" to some, the utility of NGS assays can be enormous when built on a strong foundation. The foundation of sample prep will readily prove the validity of "garbage in, garbage out". Examples of sample prep issues from commercially available NGS tests will be introduced.

11:00 Optimized Sample Handling and Target Enrichment Strategies for Challenging Clinical Samples

Bill Biggs, Ph.D., Director, Clinical Sequencing, Broad Institute of MIT and Harvard

The desire to access valuable clinical FFPE samples using advanced molecular techniques such as next-generation sequencing methods in an efficient and productive manner represents an ongoing challenge for most clinical laboratories. At the Clinical Research Sequencing Platform (CRSP) within the Broad Institute methods have been developed, optimized and implemented which allow for the ready and routine access of FFPE samples for such NGS-based analyses as Whole Exome Sequencing and Targeted Re-sequencing. These processes and methods allow for exceptional data to be obtained from even the most challenging of FFPE samples where low yield (≤ 100ng) or age (> 5 yrs) can confound analytical processes.

11:30 Automation of Sample Preparation for Clinical NGS: The Requirements and the Challenges Presented by the Various Clinical Sample Types

Martin Siaw, Ph.D., Staff Scientist and Associate Director, Advanced Sequencing Core, Advanced Sequencing, Quest Diagnostics Nichols Institute

Sample preparation is an important component of any molecular testing that is being done in clinical laboratories. With the increasing use of NGS for clinical testing comes the need to process increasingly larger numbers of patient samples. Automation of sample preparation should be considered to be critical to the workflow of diagnostic tests involving the use of NGS. My presentation will focus on the requirements and challenges for CLIA-certified clinical laboratories.

12:00 pm Better Annotation Needed at Various Points in the Sequencing Process to Avoid Reporting Errors due to Pseudogenes and High Repeat Genomic Regions

Patricia Mueller, Ph.D., Chief, Molecular Risk Assessment Laboratory, Newborn Screening and Molecular Biology Branch, DLS, Centers for Disease Control and Prevention (CDC)

Outside of clinical genomic sequencing centers, there is a general lack of awareness of the problems in NGS caused by the large number of pseudogenes in the human genome. Current estimates exceed 19,000 pseudogenes. Current standard enrichment methods do not reliably distinguish between highly homologous functional and pseudogenes. Better annotation is needed at different points in the sequencing process, including reference sequences, hybrid-capture arrays, PCR primer design software, and data-analysis software to help those designing tests and analyzing data to avoid reporting errors.

12:30 Session Break

12:40 Seven Steps to the Sample Life: Best Practices for Clinical Trial Sample Management

Sponsored by BioFortis

Mark A. Collins, Ph.D., Director, Marketing, BioFortis, Inc.

The increased interest in biomarker-based studies necessitates a new rigor and sophistication in sample and sample related data management within the clinical trial context. In addition many trials occur across geographies, are increasingly externalized with multiple stakeholders and generate large amounts of data, which is putting existing software systems, infrastructures and processes under considerable pressure. Using case studies, attendees will learn emerging best practices for clinical trial sample and data management.

1:10 Luncheon Presentation II *(Sponsorship Opportunity Available)*

1:40 Refreshment Break in the Exhibit Hall with Poster Viewing

DATA STORAGE AND MAINTENANCE

2:15 Chairperson's Remarks

Dave Anstey, Global Head, Life Sciences, YarcData

2:20 Implementing Big Data Analysis and Archival Solutions for NGS Data

*Zhiyan Fu, Ph.D., Chief Scientific Computing Officer, Genome Institute of Singapore (A*STAR)*

This presentation shows the latest development in big data analysis, compression and storage management. It provides a practical case to implement the big data technologies to a mid-size genome center. Attendees will understand the challenges of big data life-cycle management in a genome center and see how the latest big data technologies are implemented, and the pros and cons of some of the techniques, including Hadoop, HDF5, and different NGS compression algorithms evaluated by GIS.

2:50 Annotation of a Massive Dataset of Whole Genome Sequences Using a Hybrid Approach

Gerry Higgins, M.D., Ph.D., Vice President, Pharmacogenomic Science, AssureRx Health, Inc.

A collection of 17,131 whole genome sequences generated by 2nd generation sequencing using Illumina, Complete Genomics, Inc. and SOliD have been analyzed using a hybrid approach. Annotation was performed using comparative genomics modeling; sequence context; functional predictions using bioinformatics tools; epigenomic alterations; gene modeling; coding disruptions; and a supervised learning machine trained on surrogate phenotypes. Using a hybrid annotation approach lead to the discovery of novel pharmacogenomic variants that could be of value for clinical pharmacogenomics.

3:20 Semantic Technologies Offer Great Promise – with Constraints. We will Discuss a New Approach for Accelerating Research: Graph Analytics at Scale

Sponsored by YarcData

Dave Anstey, Global Head, Life Sciences, YarcData

Our platform for real-time data discovery is enabling leading research hospitals and life sciences organizations to analyze ALL their diverse data sets together, without sampling to rapidly validate more hypotheses, identify unknown relationships and get more value from their data.

3:50 Speaker to be Announced

4:20 Valentine's Day Celebration in the Exhibit Hall with Poster Viewing

5:20 Breakout Discussions in the Exhibit Hall *(see website for details)*

6:30 Close of Day

WEDNESDAY, FEBRUARY 12

7:00 am Breakfast Presentation *(Sponsorship Opportunity Available)* **or Morning Coffee**

» 8:00 PLENARY KEYNOTE SESSION
(please see page 4 for details)

9:45 Refreshment Break and Poster Competition Winner Announced in the Exhibit Hall

CLINICAL SEQUENCING
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CLINICAL SEQUENCING

continued

DIFFERENT APPROACHES TO DIAGNOSING USING NGS

10:35 Chairperson's Remarks

Karl Voelkerding, M.D., Associate Professor, Pathology, University of Utah; Medical Director, Advanced Technology & Bioinformatics, ARUP Laboratories

10:40 Use of Exome Sequencing for Genetic Diagnosis: Clinical Experience and Case Examples

Wayne W. Grody, M.D., Ph.D., Professor, Medical Genetics and Molecular Pathology, Pathology & Lab Medicine, Pediatrics, and Human Genetics; Director, Molecular Diagnostic Laboratories and Clinical Genomics Center, UCLA School of Medicine

The advent of massively parallel or "next-generation" DNA sequencing has finally brought into reach the long-anticipated "Thousand Dollar Genome", or the ability to sequence an individual's entire genome at reasonable cost. This presentation will review such aspects as clinical utility, challenges in test interpretation and genetic counseling, return of incidental findings and reimbursement, all within the context of our own experience performing clinical whole-exome sequencing at an academic medical center.

11:10 The Art of Interpreting and Reporting Results from a Multi-Gene, NGS-Based Panel for Solid Tumor Mutation Testing

Allie Grossmann, M.D., Ph.D., Staff Pathologist, Surgical Pathology and Oncology, ARUP Laboratories

With the advent of clinical testing of tumors with NGS technology, the breadth of sequence coverage has expanded beyond the scope of well-characterized mutations. This presents a challenge to both interpreting variants and to meeting reasonable clinical turn-around times for reporting. These challenges will be discussed within the context of clinical case examples with an emphasis on "lessons learned" in the adoption of NGS for solid tumor mutation testing.

11:40 Providing More Comprehensive Genetic Diagnostics by Next-Generation Sequencing-Based Multi-Gene Panels

Karl Voelkerding, M.D., Associate Professor, Pathology, University of Utah; Medical Director, Advanced Technology & Bioinformatics, ARUP Laboratories

For many genetic disorders, heterogeneity is the rule and NGS has provided a new avenue by which to overcome the limitations of Sanger sequencing-based diagnostic approaches. This presentation will highlight how NGS is transforming the diagnostic evaluation of genetic disorders through more comprehensive multi-gene panel-based methods. Technical options and bioinformatics considerations will be discussed along with how Sanger sequencing and NGS are being used in an integrative fashion.

Sponsored by
SimulConsult

12:10 pm Pertinence Metric Enables Hypothesis-Independent Genome-Phenome Analysis in Seconds

Michael M. Segal, M.D., Ph.D., Chief Scientist, SimulConsult

Genome-phenome analysis uses a genomic variant table and compares patient's findings to those of known diseases ("phenome"). Accuracy was 100% with trios with family-aware calling, and close to that with only probands. The gene pertinence metric calculated in the analysis was 99.9% for the causal genes, and the analysis took seconds and was hypothesis-independent.

Sponsored by
WAFERGEN BIOSYSTEMS

12:25 Luncheon Presentation: Sequencer-Ready Target Enrichment for Clinical Next Generation Sequencing: A Massively Parallel Singleplex PCR Approach

Xun Xu, Ph.D., Deputy Director, BGI Research, WaferGen BioSystems

Advances in NGS technology have resulted in dramatic improvements in sequencing throughput and turnaround time, yet a critical bottle neck in NGS workflows exists at the library preparation stage. We present a technology that dramatically reduces process time by generating sequencer-ready amplicon libraries in a single step. This coupled with the ability to simultaneously process multiple samples enables a high fidelity, scalable and cost effective high throughput solution for NGS targeted resequencing.

1:00 Refreshment Break in the Exhibit Hall and Last Chance for Poster Viewing

INSIGHTS INTO BIOLOGY OF CANCER FROM NGS

1:40 Chairperson's Remarks

German Pihan, M.D., Director, Hematopathology Lab, Pathology, Beth Israel Deaconess Medical Center & Harvard Medical School

1:45 The Spectrum of Mutations and Mutated Cancer Genes Across Many Tumor Types

Michael Lawrence, Ph.D., Computational Biologist, Cancer Genome Computational Analysis, Broad Institute of MIT and Harvard University

Comprehensive knowledge of the genes underlying human cancers is a critical foundation for cancer diagnostics, therapeutics, clinical trial design and selection of rational combination therapies. While some cancer genes are mutated at very high frequency (>20% of patients), the vast majority are found at intermediate frequencies (2-20%) and sometimes even lower frequencies.

2:15 The Evolving Genome of Glioblastoma

Siyuan Zheng, Ph.D., Postdoctoral Fellow, Bioinformatics & Computational Biology, MD Anderson Cancer Center, University of Texas

Glioblastoma (GBM) rates amongst the most deadly of adult tumors. We have performed extensive genomic profiling to characterize the landscape of somatic alterations of GBM. Analysis of matched pre- and post-treatment GBM showed that the GBM genome evolves under the selective pressures from cytotoxic therapy and cytoreductive surgery. We provide new insights into the heterogeneity of this disease, and the factors contributing to it.

2:45 Diffuse Large B-Cell Lymphoma

Ryan D. Morin, Ph.D., Assistant Professor, Bioinformatics, Molecular Biology and Biochemistry, Simon Fraser University

3:15 Sponsored Presentations *(Opportunities Available)*

3:45 Refreshment Break

CLINICAL-GRADE SEQUENCING AND DATA ANALYSIS

4:00 Chairperson's Remarks

4:05 CLIA-Certified Cancer Genome Diagnostic Testing in a Rapidly Changing Environment

David Wheeler, Ph.D., Co-Director, Bioinformatics; Associate Professor, Department of Molecular and Human Genetics, Baylor College of Medicine

A CLIA lab seeks reproducibility and stability in the assays performed. However, the rapidly accumulation of novel and exciting cancer genome discoveries is expanding our view of the genetic landscape of cancer and impacting how diagnostic testing can be performed. Here I will discuss analytical and biological advances that are changing the way we think about diagnostic services, and how we are integrating them into the CLIA lab.

4:35 Implementation of Next-Generation Sequencing in the Clinical Molecular Oncology Laboratory

Eric Duncavage, M.D., Assistant Professor; Director, Molecular Genetic Pathology Training Program, Department of Pathology and Immunology, Division of Clinical and Genomic Medicine, Washington University

This talk will focus on the design, informatics, and validation of clinical grade NGS-based oncology testing based on experience in the Washington University Genomics and Pathology Services Laboratory. Design considerations including enrichment methods and selecting the best genes for an oncology panel will be discussed and an evaluation of clinical-grade variant analysis tools will be provided. Finally, NGS validation practices, regulatory guidelines, and reimbursement issues will be reviewed.

5:05 Interpreting My DTC Exomes Using Public Access Clinical Databases

Gabe Rudy, Vice President, Product Development, Golden Helix

23andMe provided through a limited pilot the delivery of uninterpreted exomes in 2012. In this talk, I use GenomeBrowse and publicly available clinical databases such as ClinVar and OMIM, as well as many other integrative genomic annotations, to interpret the exomes of myself, wife and son. Recent improvements in alignment, variant calling and public annotation sources may be able to take my research grade exome to a clinical grade interpretation.

5:35 Close of Conference Program

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CANCER CHANNEL

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Cambridge Healthtech Institute's Second Annual

PREDICTIVE PRECLINICAL MODELS IN ONCOLOGY

Delivering Reproducible and Predictive Research Results

MONDAY, FEBRUARY 10

10:30 am Conference Program Registration

TUMOR MODELS AS A TOOL IN TRANSLATIONAL CANCER RESEARCH

11:50 Chairperson's Opening Remarks
Neal Goodwin, Ph.D., Vice President, Corporate Research Development, Champions Oncology, Inc.

» KEYNOTE PRESENTATIONS:

12:00 pm Modeling Efficacy and Resistance of Hedgehog Pathway Inhibitors in Cancer
Frederic de Sauvage, Ph.D., Vice President, Molecular Biology, Genentech
Hh Pathway Inhibitors have shown strong activity in cancers driven by Hh pathway mutations but have so far failed to demonstrate benefit in tumors where the pathway is activated through Hh ligand overexpression. Mouse models of cancers provide valuable tools to study the preclinical activity of Hedgehog Pathway Inhibitors and characterize potential mechanisms of innate or acquired resistance.

12:30 Bridging Tumor Genomics to Therapeutics through Patient Derived Xenografts (PDXs)
David Gandara, M.D., Professor of Medicine, Division of Hematology/Oncology, University of California, Davis School of Medicine; Director, Thoracic Oncology Program, Senior Advisor to the Director, UC Davis Comprehensive Cancer Center; Chair, Lung Committee, Southwest Oncology Group (SWOG)

1:00 Session Break

1:15 Luncheon Presentation I: The 'Innovation Universe' at Oncotest
Sponsored by oncotest
Julia Schüler, D.V.M., Ph.D., Head, Tumor Biology, Oncotest GmbH
Oncotest, a leading CRO in preclinical profiling of anti-cancer agents, has a wide range of R&D projects: Expanding our panel of PDXs, high-content/high-throughput ex-vivo assays and advanced xenograft models. These projects are often supported by collaborations with clinicians, academics and pharma partners.

1:45 Luncheon Presentation II
Sponsored by Molecular Imaging
W.R. Leopold, Ph.D., Vice President of Oncology, Molecular Imaging, Inc.

2:15 Session Break

CANCER BIOLOGY ADVANCES TO TRANSLATE INTO ACTIONABLE FINDINGS

2:30 Chairperson's Remarks
Jean-François Mirjolet, Ph.D., Technology Director, Oncodesign

2:35 Microenvironmental Regulation of GBM Progression and Therapeutic Resistance
Gabriele Bergers, Ph.D., Professor of Neurological Surgery, Neill H. and Linda S. Brownstein Endowed Chair in Brain Tumor Research, Principal Investigator, Brain Tumor Research Center, University of California, San Francisco

3:05 Resistance to MAPK Pathway Inhibitors in Melanoma: Insights and Future Challenges
Jessie Villanueva, Ph.D., Assistant Professor, Molecular & Cellular Oncogenesis Program, Melanoma Research Center, The Wistar Institute
The MAPK pathway is a key therapeutic target for melanoma as it is activated in most tumors. Despite the clinical success of drugs targeting this pathway, their therapeutic efficacy is limited by the development of drug resistance. To develop effective therapies for melanoma, it is critical to uncover the mechanisms of resistance to BRAF and MEK inhibitors. Recent studies on the molecular mechanisms of resistance to inhibitors of the MAPK pathway and potential strategies to overcome resistance will be discussed.

3:35 PANEL DISCUSSION: Translational Oncology Starts from Predictive Preclinical Models
Panelists: Speakers of the Day

4:05 Tumorgraft Models to Guide Oncology Drug Development
Sponsored by CHAMPIONS ONCOLOGY
Neal Goodwin, Ph.D., Vice President, Corporate Research Development, Champions Oncology, Inc.

4:35 Using Populations of Targeted PDX Models to Support Preclinical Trials of Oncology Therapeutics
Sponsored by MOLECULAR RESPONSE
Thomas B. Broudy, Ph.D., CSO, Molecular Response

4:50 Refreshment Break and Transition to Plenary Keynote

» 5:00 PLENARY KEYNOTE SESSION
(please see page 4 for details)

6:15 Grand Opening Reception in the Exhibit Hall with Poster Viewing

7:45 Close of Day

TUESDAY, FEBRUARY 11

7:00 am Registration and Morning Coffee

» 8:00 PLENARY KEYNOTE SESSION
(please see page 4 for details)

9:15 Refreshment Break in the Exhibit Hall with Poster Viewing
Sponsored by BIOTECHNOLOGY EXECUTIVE SEARCH

ASSURING PREDICTABILITY OF TUMOR MODELS

10:25 Chairperson's Remarks
Jocelyn Holash, Ph.D., Senior Research Investigator, Novartis

10:30 Pfizer's Strategy for the Application of Preclinical Models to Fuel Discovery Biology and Drug Discovery
Peter Olson, Ph.D., Principal Scientist, Pfizer Pharmaceuticals
Preclinical models play increasingly pivotal roles in anticancer drug development, from target identification, to candidate selection and beyond. These disease models should ideally reflect the complexity and heterogeneity of human cancer, while retaining technical features compatible with industry-level testing scale and timelines. This presentation will focus on Pfizer's strategy regarding the use of patient-derived xenografts as relevant preclinical models for cancer drug discovery.

PREDICTIVE PRECLINICAL MODELS IN ONCOLOGY
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PREDICTIVE PRECLINICAL MODELS IN ONCOLOGY

continued

11:00 Biomarker-Guided Breast Cancer Drug Evaluation in I-SPY 2

Laura J. van 't Veer, Ph.D., Professor of Laboratory Medicine, University of California San Francisco; Leader, Breast Oncology Program; Associate Director, Applied Genomics, UCSF Helen Diller Family Comprehensive Cancer Center; Principal Investigator, Bay Area Breast Cancer SPORE; Angela and Shu Kai Chan Endowed Chair in Cancer Research

11:30 Preclinical Models to Help Identify Responder Populations in the Clinic

Jocelyn Holash, Ph.D., Senior Research Investigator, Novartis

Novartis has built a range of tools that allow us to define the characteristics of cancers that are most responsive to specific targeted therapies. In this presentation I will describe these tools and provide examples of how we have used them to support clinical trials.

12:00 pm Found in Translation: Patient-Derived Xenograft Models Define Molecular Signatures of Metastasis and Therapeutic Targets

Fredika M. Robertson, Ph.D., Professor, Department of Experimental Therapeutics, Center for Targeted Therapy; Director, Translational Therapeutics Laboratory, University of Texas MD Anderson Cancer Center

This presentation will describe development of novel patient-derived xenograft models using breast tumor cells from patients with metastatic disease. Genomic pathway analysis identified enrichment of genes that regulate maintenance of a cancer stem cell phenotype and those that regulate hypoxia, glucose uptake and glutamine metabolism. These insights into pathways activated during breast tumor progression can be matched by existing and emerging therapies for development of clinical trials.

12:30 Session Break

Sponsored by Oncodesign

12:40 Luncheon Presentation I: Creating a Functional Human Immune System in Mice to Better Predict Efficacy of New Classes of Oncology and Immunology Drugs

Jean-François Mirjolet, Ph.D., Technology Director, Oncodesign

Tumor and inflammation microenvironments are the targets of new classes of therapeutic drugs. Second generation NOG mice are now available that surpass the limitations of the previous generation. For example, mice expressing human IL2 improve *in vivo* human NK functionality and mice expressing human HLA allow for the *in vivo* evaluation of human HLA restricted function.

1:10 Luncheon Presentation II *(Sponsorship Opportunity Available)*

1:40 Refreshment Break in the Exhibit Hall with Poster Viewing

2:15 Chairperson's Remarks

Serguei Kozlov, Ph.D., Principal Scientist, Center for Advanced Preclinical Research, SAIC-Frederick, Inc.

2:20 PREDECT in Europe: Goals and Outcomes

Ralph Graeser, Ph.D., Associate Director, Janssen Pharmaceutica NV

PREDECT is an IMI-funded partnership between 9 academic, 3 SME and 9 EU pharmaceutical company partners, developing advanced, transferable *in vitro* models for breast, prostate and lung cancers.

2:50 Increasing Predictability of Tumor Animal Models via Systematic Validation

Terry A. Van Dyke, Ph.D., Head, Mouse Cancer Genetics Program; Program Director, Cancer Pathways and Mechanisms, National Cancer Institute

Sponsored by The Jackson Laboratory

3:20 Preclinical and Clinical Applications of Patient Derived Xenograft (PDX) Models

Yan Yang, Ph.D., Director, Laboratory Operations, In Vivo Services The Jackson Laboratory

We have established a unique collaboration with over 20 clinical centers to advance cancer treatment. Patient tumors transplanted into the NSG mouse are being screened with SOC and experimental therapeutics for preclinical research or the refinement of patient treatment regimens.

3:50 PANEL DISCUSSION: Challenging Cancer Heterogeneity with Reproducible and Predictive Preclinical Studies

Moderator: Terry A. Van Dyke, Ph.D., Head, Mouse Cancer Genetics Program; Program Director, Cancer Pathways and Mechanisms, National Cancer Institute

Panelists: Speakers of the Session

4:20 Valentine's Day Celebration in the Exhibit Hall with Poster Viewing

5:20 Breakout Discussions in the Exhibit Hall *(see website for details)*

6:30 Close of Day

WEDNESDAY, FEBRUARY 12

7:00 am Breakfast Presentation *(Sponsorship Opportunity Available)* **or Morning Coffee**

8:00 PLENARY KEYNOTE SESSION *(please see page 4 for details)*

9:45 Refreshment Break and Poster Competition Winner Announced in the Exhibit Hall

MODELS FOR CANCER IMMUNOTHERAPY

10:35 Chairperson's Remarks

Tom Metcalfe, BSc, MBA, Managing Director, Oncotest

10:40 Targeting Cancer with BiTE™ Molecules

Holger Wesche, Ph.D., Scientific Director, Oncology, Amgen, Inc.

The recent clinical success in harnessing the immune system to fight cancer has demonstrated the potential of this approach. Here we will give an overview of the BiTE (Bi-specific T-cell Engager) technology, which drives directed T cell mediated tumor cell killing.

11:10 Molecular Analysis of Mouse Syngeneic Tumor Models as a Translational Tool in Understanding Anti-PD1 Immunotherapy

Terrill K. McClanahan, Ph.D., Senior Principal Scientist, Molecular Discovery, Biology & Pharmacology Merck Research Labs, Biologics

The utility of mouse syngeneic tumor models to develop immunomodulatory agents for oncology will be discussed. We have studied anti-tumor efficacy, pharmacology and pharmacodynamics of a surrogate anti-mouse PD-1 antibody for mechanistic biomarker discovery and translation to human clinical studies.

11:40 Missing The Targets: Why *In Vivo* Models Are Not Up To The Job

Bent Jakobsen, Ph.D., CSO, Immunocore

Since immunotherapies interact specifically with human target molecules, preclinical assessment using animal models requires careful consideration. We describe an alternative approach based on *in vitro* molecular and cellular testing, and apply this strategy to a novel TCR-based therapeutic possessing dual specificity for human targets.

12:10 pm Session Break

12:20 Luncheon Presentation *(Sponsorship Opportunity Available)* **or Lunch on Your Own**

1:00 Refreshment Break in the Exhibit Hall and Last Chance for Poster Viewing

PREDICTIVE PRECLINICAL MODELS IN ONCOLOGY
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PREDICTIVE PRECLINICAL MODELS IN ONCOLOGY

continued

CHALLENGING CANCER GENOMIC HETEROGENEITY

1:40 Chairperson's Remarks
Vivienne Watson, Ph.D., Senior Scientist, Amgen

1:45 Modeling Tumor Heterogeneity: Accounting for CSCs and the Immune System
Elaine Hurt, Ph.D., Scientist II, MedImmune
At MedImmune we are developing *in vivo* models encompassing several important attributes of cancer, including recapitulating the heterogeneity of tumors by using patient-derived xenografts, understanding the contribution of cancer stem cells and circulating tumor cells, as well as modeling the role of the immune system. Through the development and use of these complex biological systems we strive to develop therapeutics that have long-lasting benefit to our patients.

2:15 Integration of Cell Line Sensitivity and Genomic Profiling Data for Patient Stratification
Vivienne Watson, Ph.D., Senior Scientist, Amgen
We have performed a functional screen using an siRNA library representing more than 14,000 genes on a set of cancer cell lines that contain genetic lesions known to be representative of a subset of lung cancer primary tumors. An association analysis to look for genomic markers that correlate with sensitivity, and that would identify a potential target patient population, was carried out. The data will be discussed.

2:45 Paradoxical Acceleration of Pancreatic Tumorigenesis through Smoothened Inhibition: A Post-Clinical Trial in Genetically Engineered Mice
Kenneth Olive, M.D., Assistant Professor of Medicine and Pathology, Irving Cancer Research Center, Columbia University
Numerous agents are being developed to attack the stroma of pancreatic tumors, despite the failure of one such agent, a Smoothened inhibitor, in clinical trials. We performed a post-clinical evaluation of the consequences of Smoothened inhibition and found that acute versus chronic dosing regimens had opposite effects of overall survival. The mechanistic basis for these difference are under investigation.

3:15 Sponsored Presentations *(Opportunities Available)*

3:45 Refreshment Break

OPTIMIZING TUMOR MODELS

4:00 Chairperson's Remarks
William J. Murphy, Ph.D., Professor and Acting Chair, Department of Dermatology; Professor, Department of Internal Medicine, Division of Hematology/Oncology, University of California, Davis School of Medicine

4:05 Development of a Clinically Relevant Model of Metastatic Colorectal Cancer
Kevin G. Leong, Ph.D., Scientist, Discovery Oncology, Genentech, Inc.
Despite the availability of xenograft, chemical-induced, and genetically-engineered mouse models of colorectal cancer, none of these models reproducibly metastasize to target organs relevant to the human disease. We have developed a novel mouse model of metastatic colorectal cancer that exhibits robust metastasis to clinically relevant target organs, and have utilized this model to investigate routes of metastatic dissemination.

4:35 Impact of Aging and Body Fat on Outcome in Preclinical Cancer Models
William J. Murphy, Ph.D., Professor and Acting Chair, Department of Dermatology; Professor, Department of Internal Medicine, Division of Hematology/Oncology, University of California, Davis School of Medicine
We have found that acute toxicities can be observed when using older but not young mice and this is dependent on body fat and a heightened inflammatory state following immunotherapies.

5:05 Increasing the Breadth and the Bandwidth of Preclinical Assessment in Biologically Engineered Murine Cancer Models
Serguei Kozlov, Ph.D., Principal Scientist, Center for Advanced Preclinical Research, SAIC-Frederick, Inc.
Genetic *de novo* cancer models have been earning rising attention as capable of providing experimental platforms featuring autochthonous carcinogenesis for informative decisions at preclinical stages. These models however are laborious, costly and require versatile expertise to manage workflows involving mouse lines engineered to mimic cancer. Strategies have been developed to "retool" conventional GEMs into more tractable alternatives to broaden and accelerate their translational applications.

5:35 Close of Conference Program

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Each year, the Informatics Channel at the Molecular Medicine Tri-Conference brings together leading experts in drug discovery informatics, bioinformatics, and IT. Through lectures, panel discussions, and interactive breakouts, we explore cutting-edge ways to manage, analyze, and integrate data to enable drug discovery, biology, and clinical research.

- Bioinformatics for Big Data
- Integrated R&D Informatics & Knowledge Management
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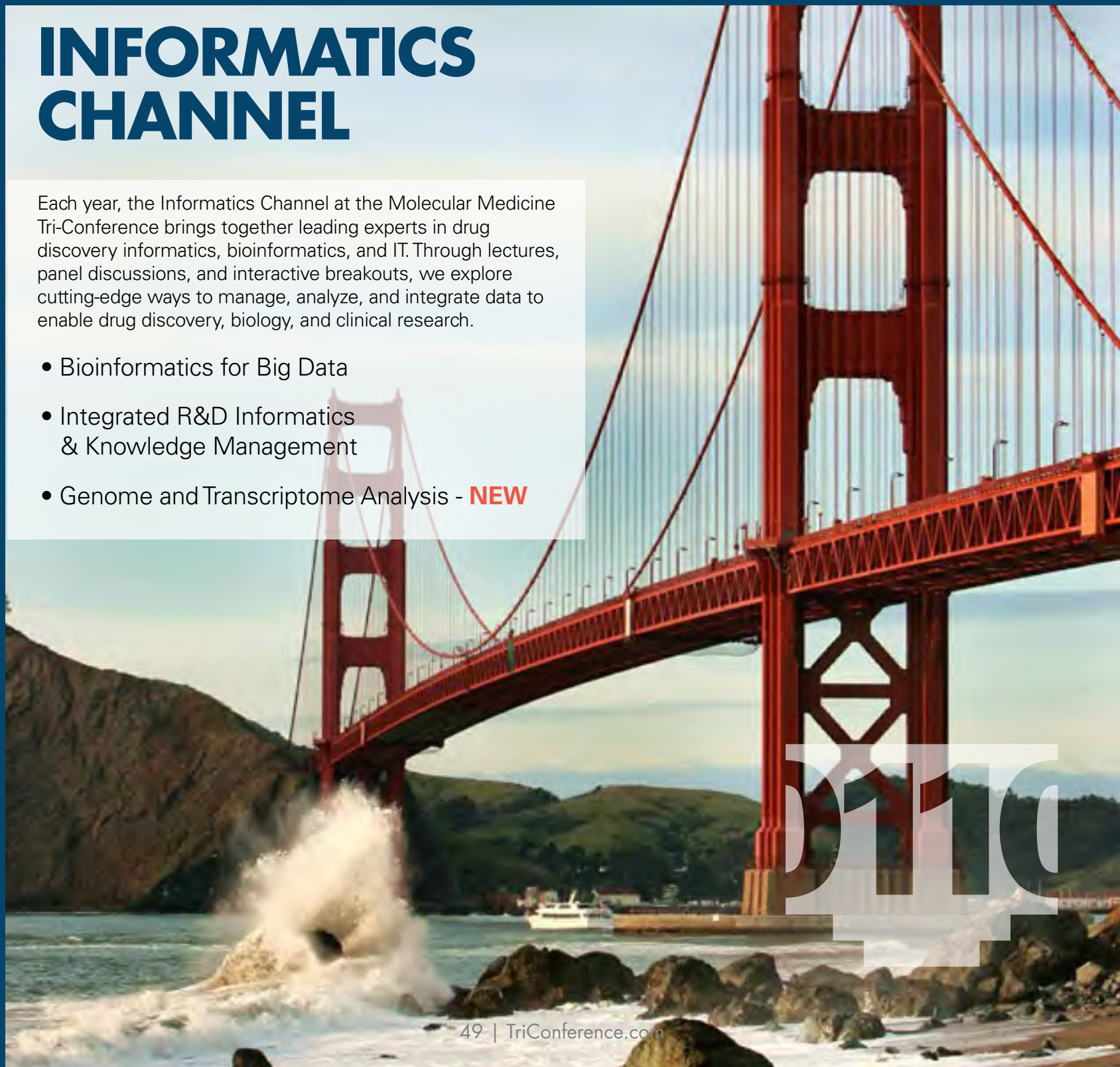

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BIOINFORMATICS FOR BIG DATA

How Applications of Big Data will Drive Research Forward

MONDAY, FEBRUARY 10

10:30 am Conference Program Registration

MOVING SYSTEMS AND BIOINFORMATICS TOWARDS PERSONALIZED MEDICINE

11:50 Chairperson's Opening Remarks

» KEYNOTE PRESENTATION:

12:00 pm Customized Care in Moving Systems and Bioinformatics towards Personalized Medicine
Richard Kellner, Co-founder and CEO, Genome Health Solutions

» 12:30 FEATURED PRESENTATION:

Crohnology: The Patient-Powered Research Network
Sean Ahrens, Co-Founder, Crohnology
Crohnology enables any patient to contribute to research for their condition and cure. Crohnology now serves over 4,400 patients with Crohn's & Colitis, and is partnered with top non-profits and industry researchers to advance treatments and the cure. Crohnology is enabling the crowd-running of clinical studies, powered by the most underutilized resource in medicine, patients themselves, and started by myself, a patient with Crohn's since age 12. This talk shares data collection methods, tools, and applications.

1:00 Session Break

1:15 Luncheon Presentations *(Sponsorship Opportunities Available)* **or Lunch on Your Own**

2:15 Session Break

WHY MEDICAL RESEARCH DOES BIG DATA WRONG

2:30 Chairperson's Remarks
Michael Liebman, Ph.D., Managing Director, Strategic Medicine, Inc.
This session brings together thought leaders from academia, government, healthcare and biopharma to discuss current big data strategies and why there is too much emphasis on genomic data collection as a driver for big data. Speakers will give a presentation and then convene for a panel discussion.

2:35 Understanding the Needs of a Healthcare System in Data Acquisition and Data Analysis
Hal Wolf, Senior Vice President (ret), COO, Permanente Federation, Kaiser Permanente

3:05 Prioritization of Big Data for Pharmaceutical Development: What and Why
Charlie Barr, M.D., Ph.D., Group Medical Director and Head, Evidence Science and Innovation, Genentech Corp.

3:35 Applying Big Data in the Clinic: What Data Do We Need and How Would We Use It?
Michael Liebman, Ph.D., Managing Director, Strategic Medicine, Inc.
Sabrina Molinaro, Ph.D., Institute for Clinical Physiology, National Research Council, Italy

4:05 Accelerating Research by Enabling Big Data in the Cloud
Sponsored by aspera
Michelle Munson, President & CEO, Aspera
Discover the technologies that enable big data to be securely transported, shared and stored in the cloud. Attendees will learn from the experiences of organizations that deployed cloud-based research platforms based on high-speed data transport, and the benefits they've achieved in terms of increased collaboration, accelerated research and reduced costs.

4:35 Fusion Genes Uncovered - Bioinformatics in Action
Sponsored by MediSapiens
Kalle Ojala, Chief Product Officer, MediSapiens Ltd.
Gene fusion are known to be pivotal events in cancer and important drug development targets. A recent example of this is Crizotinib targeting the EML4-ALK fusion in non-small cell lung cancer. We have developed a new bioinformatic tool that allows efficient identification of fusion genes in large RNA sequencing datasets. Our tool allows for the efficient analysis of impact of the gene fusion as well its clinical significance.

4:50 Refreshment Break and Transition to Plenary Keynote

» 5:00 PLENARY KEYNOTE SESSION
(please see page 4 for details)

6:15 Grand Opening Reception in the Exhibit Hall with Poster Viewing

7:45 Close of Day

TUESDAY, FEBRUARY 11

7:00 am Registration and Morning Coffee

» 8:00 PLENARY KEYNOTE SESSION
(please see page 4 for details)

9:15 Refreshment Break in the Exhibit Hall with Poster Viewing
Sponsored by BIOTAL PARTNERS EXECUTIVE SEARCH

TECHNOLOGIES GENERATING BIG BIOMEDICAL DATA

10:25 Chairperson's Remarks
Martin Gollery, CEO, Tahoe Informatics

10:30 Breaking Down the Wave: A Look at the Data Sources that are Transforming Research
Martin Gollery, CEO, Tahoe Informatics
For well over a decade, the amount of biological data has grown at a rate that exceeds Moore's Law, a phenomenon that is commonly compared to a 'Tsunami' of data. Today that acceleration continues, with the added complexity from a wide range of disparate data sources that must be integrated and filtered by researchers to build a coherent picture of the system being studied. This talk will be a high-level overview of the different technologies that generate big data in the biomedical arena. Finally, we will look into the future at upcoming technologies and the challenges and opportunities that they will present.

11:00 Cancer Genomics
David Haussler, Ph.D., Distinguished Professor and Director, Center for Biomolecular Science & Engineering, University of California Santa Cruz
UCSC has built the Cancer Genomics Hub (CGHub) for the US National Cancer Institute, designed to hold up to 5 petabytes of research genomics data (up to 50,000 whole genomes), including data for all major NCI projects. To date it has served more than 8.3 petabytes of data to more than 300 research labs. Cancer is exceedingly complex, with thousands of subtypes involving an immense number of different combinations of mutations. The only way we will understand it is to gather together DNA data from many thousands of cancer genomes so that we have the statistical power to

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distinguish between recurring combinations of mutations that drive cancer progression and "passenger" mutations that occur by random chance. Currently, with the exception of a few projects such as ICGC and TCGA, most cancer genomics research is taking place in research silos, with little opportunity for data sharing. If this trend continues, we lose an incredible opportunity. Soon cancer genome sequencing will be widespread in clinical practice, making it possible in principle to study as many as a million cancer genomes. For these data to also have impact on understanding cancer, we must begin soon to move data into a global cloud storage and computing system, and design mechanisms that allow clinical data to be used in research with appropriate patient consent. A global alliance for sharing genomic and clinical data is emerging to address this problem. This is an opportunity we cannot turn away from, but involves both social and technical challenges.

11:30 A Test for Predicting Cardiovascular Death in Coronary Artery Disease Patients

Reijo Laaksonen, M.D., Ph.D., FESC, CMO, Zora Biosciences Oy

LDL-cholesterol (LDL-C) has traditionally been used to gauge cardiovascular risk. However, LDL-C provides only limited predictive information on fatal CVD complications in patients with established coronary artery disease (CAD). These patients at the highest risk of myocardial infarction or cardiovascular death present an unmet diagnostic need. Identification of these individuals would allow their more focused treatment in time preventing pre-mature deaths and hospitalizations. In our effort to address this unmet diagnostic need we applied the Zora lipidomic technology to patient samples from well defined CAD patient cohorts. We successfully developed markers that identify high risk CAD patients with accuracy that cannot be reached with currently used routine clinical measurements. Importantly these markers are actionable and can be used also for monitoring the treatment success in patients. The Zora high risk CAD test will lead to significant improvements in clinical diagnostics with concomitant health care savings.

12:00 pm Exploring Microbiome in Metabolic Diseases

Deepak K. Rajpal, D.V.M., Ph.D., Director, Computational Biology, GlaxoSmithKline

Metabolic diseases, especially type 2 diabetes and obesity, are growing global healthcare concerns. Various studies have highlighted the role of gastrointestinal microbial communities in metabolic health and disease. We will provide a brief overview of the gut microbiome, its putative role in metabolic diseases and the emerging data in this space.

12:30 Session Break

12:40 Luncheon Presentations *(Sponsorship Opportunities Available)* **or Lunch on Your Own**

1:40 Refreshment Break in the Exhibit Hall with Poster Viewing

DATA STORAGE AND MAINTENANCE

2:15 Chairperson's Remarks

Dave Anstey, Global Head, Life Sciences, YarcData

2:20 Implementing Big Data Analysis and Archival Solutions for NGS Data

*Zhiyan Fu, Ph.D., Chief Scientific Computing Officer, Genome Institute of Singapore (A*STAR)*

This presentation shows the latest development in big data analysis, compression and storage management. It provides a practical case to implement the big data technologies to a mid-size genome center. Attendees will understand the challenges of big data life-cycle management in a genome center and see how the latest big data technologies are implemented, and the pros and cons of some of the techniques, including Hadoop, HDF5, and different NGS compression algorithms evaluated by GIS.

2:50 Annotation of a Massive Dataset of Whole Genome Sequences Using a Hybrid Approach

Gerry Higgins, M.D., Ph.D., Vice President, Pharmacogenomic Science, AssureRx Health, Inc.

A collection of 17,131 whole genome sequences generated by 2nd generation sequencing using Illumina, Complete Genomics, Inc. and SOliD have been analyzed using a hybrid approach. Annotation was performed using comparative genomics modeling; sequence context; functional predictions using bioinformatics tools; epigenomic alterations; gene modeling; coding disruptions; and a supervised learning machine trained on surrogate phenotypes. Using a hybrid annotation approach lead to the discovery of novel pharmacogenomic variants that could be of value for clinical pharmacogenomics.

Sponsored by
YarcData

3:20 Semantic Technologies Offer Great Promise – with Constraints. We will Discuss a New Approach for Accelerating Research: Graph Analytics at Scale

Dave Anstey, Global Head, Life Sciences, YarcData

Our platform for real-time data discovery is enabling leading research hospitals and life sciences organizations to analyze ALL their diverse data sets together, without sampling to rapidly validate more hypotheses, identify unknown relationships and get more value from their data.

GENOME-WIDE PROTEIN STRUCTURE AND FUNCTION PREDICTION

3:50 Genome-Wide Protein Structure and Function Prediction

Andrzej Kloczkowski, Ph.D., Professor, Battelle Center for Mathematical Medicine, The Research Institute at Nationwide Children's Hospital and Department of Pediatrics, The Ohio State University College of Medicine

The knowledge of protein structure is critical to comprehend their function, for understanding of molecular mechanisms of disease, and for development of new generations of medicines based on the computer-aided drug design. Because of this there is an urgent need to improve the existing computational methods of structure prediction to reach ultimately the accuracy of prediction comparable to crystallographic or NMR structure determination resolution. We discuss these important problems and propose new methods for genome-wide protein structure and function prediction.

4:20 Valentine's Day Celebration in the Exhibit Hall with Poster Viewing

5:20 Breakout Discussions in the Exhibit Hall *(see website for details)*

6:30 Close of Day

WEDNESDAY, FEBRUARY 12

7:00 am Breakfast Presentation *(Sponsorship Opportunity Available)* **or Morning Coffee**

8:00 PLENARY KEYNOTE SESSION
(please see page 4 for details)

9:45 Refreshment Break and Poster Competition Winner Announced in the Exhibit Hall

DATA INTEGRATION & SHARING: TOOLS AND POLICIES FOR VISUALIZATION AND ANALYSIS

10:35 Chairperson's Remarks

Larry Hunter, Ph.D., Director, Center for Computational Pharmacology & Computational Bioscience Program, Professor, Pharmacology, University of Colorado

10:40 Knowledge-Based Analysis at Genomic Scale

Larry Hunter, Ph.D., Director, Center for Computational Pharmacology & Computational Bioscience Program, Professor, Pharmacology, University of Colorado

High-throughput instruments and the explosion of new results in the scientific literature is both a blessing and a curse to the bench researcher. Effective design and implementation of computational tools that genuinely facilitate the generation of novel and significant scientific insights remains poorly understood. This talk presents efforts that combine natural language processing for information extraction, graphical network models for semantic data integration, and some novel user interface approaches into a system that has facilitated several significant discoveries.

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BIOINFORMATICS FOR BIG DATA

continued

11:10 Combining Visual Analytics and Parallel Computing for Data-Driven Analysis Pipeline Selection and Optimization to Support the Big Data to Knowledge Transformation

Richard Scheuermann, Director, Informatics, J. Craig Venter Institute

This presentation describes our efforts at the J. Craig Venter Institute (JCVI) in collaboration with the Texas Advanced Computing Center (TACC) in the development of a high performance cyber-infrastructure that combines visual analytics and parallel computing for data-driven selection and optimization of analytical pipelines based on objective performance metrics. We demonstrate the application of these principles and infrastructure for the analysis of genome-wide gene expression and high-throughput, high dimensional flow cytometry data in clinical and translational research settings.

11:40 Co-Presentation: Spotfire Templates for Analysis & Visualization of Project Data

Sandhya Sreepathy, PMP, Head of Operations, Global Discovery Chemistry, Novartis

Heather Hogg, Ph.D., Business Analyst, Novartis Institutes for Biomedical Research

There has been a significant increase in the usage of Spotfire by project teams in Emeryville for visualization and analysis of data. To help streamline development activity and support the needs of project teams with data analysis and visualization, NIBR-IT in collaboration with computational chemistry group in Emeryville developed project based Spotfire templates. Templates were built leveraging existing technologies for retrieval/import of data and incorporated common elements of analysis and visualization(scaffold assignment, ligand, lipophilic efficiency, activity ratios, r-group decomposition etc). Predefined visualizations and filters helped accelerate project team decision making.

12:10 pm Session Break

12:20 Luncheon Presentation *(Sponsorship Opportunity Available)* **or Lunch on Your Own**

1:00 Refreshment Break in the Exhibit Hall and Last Chance for Poster Viewing

HOW BIG DATA WILL DRIVE RESEARCH FORWARD

1:40 Chairperson's Remarks

Michael H. Elliott, CEO, Atrium Research & Consulting LLC

1:45 Harnessing Big Data to Accelerate Drug Development

Vinod Kumar, Ph.D., Senior Investigator, Computational Biology, GlaxoSmithKline Pharmaceuticals

With the rapid development of high-throughput technologies and ever-increasing accumulation of whole genome-level datasets, an increasing number of diseases and drugs can be comprehensively characterized by the changes they induce in gene expression, protein, metabolites and phenotypes. Integrating and querying such large volumes of data, often spanning domains and residing in diverse sources, constitutes a significant obstacle. This talk presents two distinct approaches that utilize these data types to systematically evaluate and suggest new disease indications for new and existing drugs.

2:15 Drug Process Design Improvement based on Data Management and Analysis

Valérie Vermylen, Knowledge Management, Director, GPS, UCB

Most of the scientific process data generated are not free to access, even if managed in databases. At UCB, data was recently made available including its context. It allows process developers to draw easily designed space and define critical parameters. To support investigation studies as impact analysis, manufacturing dashboards and trends are automatically published. An example of correlation between process data and patients' clinical responses will be presented as an illustration of advanced data analysis.

2:45 The Library of Integrated Network-based Cellular Signatures (LINCS) Information FramEwork (LIFE)

Stephan C. Schürer, Ph.D., Associate Professor, Pharmacology, Center for Computational Science at Miller School of Medicine, University of Miami

The NIH-funded LINCS consortium is producing an extensive dataset of cellular response signatures to a variety of small molecule and genetic perturbations. We have been developing the LINCS Information FramEwork (LIFE) – a specialized knowledge-driven search system for LINCS data.

3:15 An Enhanced Molecular Design Platform That Fosters Ideation, Knowledge Transfer, and Collaboration

Sponsored by SCHRÖDINGER

Leah Frye, Ph.D., Vice President, Drug Discovery Applications Group, Schrödinger

Drug discovery is the ultimate team sport. Schrödinger is developing a collaborative and knowledge engineered platform—LiveDesign—to help scientists not only capture their ideas and best practices, but to exploit and share these with select team members. Above and beyond the aggregation of 2D data, this platform will allow users to bring together 3D data with its associated annotations. LiveDesign will ultimately lead to better patient outcomes, promoting better scientific communication by exposing data, ideas, and colleague feedback during the design and redesign phases of molecular discovery.

3:45 Refreshment Break

BIG DATA DRIVING PERSONALIZED MEDICINE

4:00 Chairperson's Remarks

Dalia Cohen, Ph.D., Founder & President, ALN Associates

4:05 Big Data's Big Role in Understanding Complex Diseases

Andreas Kogelnik, M.D., Ph.D. Founder and Director, Open Medicine Institute

The Open Medicine Institute (OMI) is effectively applying a collaborative, "big data" approach to understand and address complex diseases including: Autism, Lyme, Chronic Fatigue Syndrome, Parkinson's, and various cancers. This presentation will discuss the creation of a patient-centric infrastructure that handles and analyzes genomic sequencing information, bio-sampling data, physiology tests, basic research, patient experiences and physician evaluations to deliver needed information about a range of diseases.

4:35 Progress on Aggregating all the World's Genetic Tests into a Single Assay

Randy Scott, Ph.D., CEO and Co-Founder, InVitae

Technology is moving rapidly to enable massively parallel genetic testing. The ability to sequence DNA, however, is only the first step in building the infrastructure to analyze, store, manage, and interpret medical genetic information for patients. InVitae is focused on building the infrastructure to bring more comprehensive genetic testing into routine medical practice throughout the world.

5:05 It's Not Just About Big Data...Big Analytics for Identifying What Works and for Whom in Healthcare

Iya Khalil, Ph.D., Executive Vice President and Co-Founder, GNS Healthcare

We are living in the era of big data in healthcare, with unprecedented ability to collect data at multiple levels (genomic/'omic', phenotypic, health records, mobile health, etc.) and at scale. The key will be leveraging advanced analytics and appropriate feedback loops to identify what works on an individual patient level.

5:35 Close of Conference Program

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INTEGRATED R&D INFORMATICS & KNOWLEDGE MANAGEMENT

Supporting Collaboration, Externalization, Globalization & Translational Research

MONDAY, FEBRUARY 10

10:30 am Conference Program Registration

EXTERNAL COLLABORATION BEST PRACTICES: GETTING VALUE OUT OF DATA WITH YOUR COLLABORATORS

11:50 Chairperson's Opening Remarks

Martin Leach, Vice President, Research & Development IT, Biogen Idec

12:00 pm Roche's Translational and Clinical Research Center (TCRC): How Our Big Data and Externalization Strategy Impacts Drug Discovery

Juergen Hammer, Ph.D., MBA, Pharma Research and Early Development Informatics (pREDi), pREDi Center Head; Global Head, Disease & Translational Informatics, Roche Translational Clinical Research Center

Pharmaceutical companies increasingly embed their research and early development organizations into vibrant academic hubs to enhance innovation and asset finding. Roche has recently opened "TCRC, Inc.", soon to be located in New York City. The presentation will focus on our Big Data and Externalization approaches to support the TCRC, and will exemplify how we impact drug development decisions using informatics.

12:30 The Lilly Open Innovation Drug Discovery Program (OIDD)

Daniel H. Robertson, Ph.D., Senior Director, LRL IT Research, Eli Lilly and Company

Through OIDD, Lilly has established a network of top global research talent at academic and biotech institutions to provide them access to proprietary, *in vitro* phenotypic, and target-based assays (PD2 and TargetD2). In addition to supplying data that may lead to potential collaborations, Lilly has recently been partnering to deploy additional design tools for OIDD investigators to assist in designing compounds submitted to the assays through this collaboration.

1:00 Session Break

1:15 Luncheon Presentation I: Building and Linking Disease and Drug Target Profiles Using Semantic Search Technologies

Sponsored by ELSEVIER

Maria Shkrob, Ph.D., Senior Bioinformatics Scientist, Elsevier

Researchers face a growing challenge in managing vast quantities of unstructured data to find relevant information that can guide their research. A new semantic search engine that incorporates text-mining capability along with customizable dictionaries and taxonomies rapidly finds facts and provides summary tables from multiple sources, including scientific abstracts and full-texts, grant applications, and in-house documents. This ability to accurately retrieve and summarize information significantly increases researcher productivity compared to traditional keyword search.

1:45 Luncheon Presentation *(Sponsorship Opportunity Available)*

2:15 Session Break

2:30 Chairperson's Remarks

Martin Leach, Vice President, Research & Development IT, Biogen Idec

2:35 Building an Informatics Ecosystem for Externalized R&D

Sándor Szalma, Ph.D., Head, External Innovation, R&D IT, Janssen Research & Development, LLC

Pharma companies have historically been involved in many partnerships fueling their discovery engines, supported with non-optimal IT systems. With recent wide-spread adaptation of hosted solutions and cloud computing, there is an opportunity now to implement informatics solutions such that collaborative exploration of the data generated in partnerships becomes possible. We also discuss the opportunities to build parts of the ecosystem in a pre-competitive manner and our experience in deploying open source tools.

3:05 Cloud Solutions Spanning Applications across R&D, Development, G&A, and Compliance Functions

John Reynders, CIO, Moderna Therapeutics; former Vice President, Research & Development Information, AstraZeneca

This presentation will overview Moderna's aggressive push into cloud solutions spanning applications across R&D, development, G&A, and compliance functions. Moderna's cloud-based informatics workflows for the design, development, screening, and delivery of messenger RNA therapeutics will be shared along with associated challenges of Big Data, cloud security, collaboration, and cross-cloud integration.

3:35 PANEL DISCUSSION: Approaches and Lessons Learned to Build a Comprehensive R&D Search Capability

Martin Leach, Vice President, Research & Development IT, Biogen Idec

The accessibility of information within any R&D organization is key to the successful collaboration and development of a research pipeline. The holy grail for most research organizations is the one-stop search (aka. Google-like search for R&D). In this panel we will discuss the approaches a number of research organizations have taken, successes, failures and lessons learned.

Panelists:

John Koch, Director, Scientific Information Architecture & Search, Merck

4:05 Efficient Data Mining for Precision Medicine

Sponsored by THOMSON REUTERS

Mark Hughes, Ph.D., Senior Product Manager, Thomson Reuters

4:35 Refreshment Break and Transition to Plenary Keynote

5:00 PLENARY KEYNOTE SESSION *(please see page 4 for details)*

6:15 Grand Opening Reception in the Exhibit Hall with Poster Viewing

7:45 Close of Day

TUESDAY, FEBRUARY 11

7:00 am Registration and Morning Coffee

8:00 PLENARY KEYNOTE SESSION *(please see page 4 for details)*

9:15 Refreshment Break in the Exhibit Hall with Poster Viewing

Sponsored by

INTEGRATED R&D INFORMATICS & KNOWLEDGE MANAGEMENT
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INTEGRATED R&D INFORMATICS & KNOWLEDGE MANAGEMENT

continued

REGISTRATION SYSTEMS

10:25 Chairperson's Remarks
Arturo J. Morales, Ph.D., Global Lead, Biology Platform Informatics, Novartis Institutes for Biomedical Research

10:30 Registration Systems: Applications or Data Stores?
Arturo J. Morales, Ph.D., Global Lead, Biology Platform Informatics, Novartis Institutes for Biomedical Research
Registration systems are not applications that usually stand on their own. Their value comes from the enablement of downstream data analysis and sample tracking through proper management of concept and sample metadata. As such, most registration systems offer little intrinsic value to those that use it directly and user compliance can be a challenge. Thus, it is important to adapt to workflows, as opposed to making users adapt to them.

11:00 Development of a LIMS Platform to Manage Biological Therapeutics
David M. Sedlock, Ph.D., Senior Director, Research & Development Systems, Takeda Cambridge US
The management of biological samples for testing as bioherapeutic agents requires a unique type of LIMS to handle both workflows and sample registration. We are currently engaged in a couple of projects at Takeda to create a working solution for both research and preclinical development samples to be managed across multiple R&D sites. The project status and business impact will be reviewed.

11:30 An Enhanced Electronic Laboratory Notebook to Support Biologics Research and Development
Beth Basham, Ph.D., Director, Account Management, Biologics Discovery & IT Site Lead, Merck
Merck is evolving our electronic lab notebook from a straightforward paper notebook replacement to a platform that structures data and results, provides basic LIMS-like capabilities and enables powerful search and analytics. We will share our experiences in providing a solution to support some of the stages of biologics research and development.

12:00 pm Biological Registration Systems at UCB and How They Integrate into the Discovery Workflow
David Lee, Ph.D., Principal Scientist, Informatics, UCB
The benefits of informatics-driven data management systems are well known in the small molecule therapeutics arena. Extending these systems to supporting biotherapeutics presents a number of challenges. We present a novel data management system, BioQuest, integrating bespoke and best in class software systems designed to capture and integrate NBE data at UCB. We will focus on registration systems, in particular on the antibody and non-antibody protein registration system based on the Genedata Biologics platform.

12:30 Session Break

Sponsored by CAMBRIDGE SEMANTICS

12:40 Luncheon Presentation I Semantics for Rapid Development of Informatics Solutions
Ben Szekely, Director & Founding Engineer, Cambridge Semantics
R&D Informatics present a demand for huge quantities of dispersed and diverse data coupled with constantly shifting regulatory and competitive landscape. In this talk, we will discuss how semantics enables - flexible conceptual information modeling; integrating structured and unstructured data at conceptual level; varied data access pathways including blending structured search, semantic search, key word search, and chemical search; and sophisticated text analytics for literature, patents, and other unstructured data sources

1:10 Luncheon Presentation II *(Sponsorship Opportunity Available)*

1:40 Refreshment Break in the Exhibit Hall with Poster Viewing

PRE-COMPETITIVE COLLABORATION & SUPPORTING PUBLIC-PRIVATE PARTNERSHIPS

2:15 Chairperson's Remarks
Barry Bunin, Ph.D., CEO, Collaborative Drug Discovery (CDD, Inc.)

Sponsored by CDD VAULT

2:20 Modern Drug Research Informatics Applications to CNS, Infectious, Neglected, Rare, and Commercial Diseases
Barry Bunin, Ph.D., CEO, Collaborative Drug Discovery (CDD, Inc.)
There are currently hundreds of commodity technologies for handling scientific information – each with its own scope and limitations. The application of collaborative technologies to interrogate potency, selectively, and therapeutic windows of small molecule structure activity relationship (SAR) data will be presented in 5 case studies. Given external (public and collaborative) data grows faster than internal data, novel collaborative technologies to gracefully manage combined external and private data provide an ever-increasing competitive advantage.

2:50 tranSMART: Use Cases from Deployments Highlighting Emerging Models for Pre-Competitive Collaboration and Open Source Sustainability
Asif Dhar, M.D., CMIO & Principal, Deloitte Consulting
The tranSMART open source translational research knowledge management software continues to make forward progress since the initial release in 2012. Specific use cases from a variety of projects incorporating tranSMART will be walked through to highlight emerging pre-competitive collaboration models including opportunities, new capabilities, and unresolved challenges. The current state of open project sustainability and approaches taken by Recombinant and other groups to ensure the software increases in value for adopters will be explored.

3:20 The Innovative Medicines Initiative: Collaborating around Knowledge Management
Anthony Rowe, Ph.D., Principal Scientist, External Innovation, Johnson & Johnson
The Innovative Medicines Initiative (IMI) is a public-private partnership between the European Federation of Pharmaceutical Industry and Associates (EFPIA) and the European Union. It is dedicated to overcoming key bottlenecks in pharmaceutical research by enabling pre-competitive collaboration between industry and academic scientists. In this talk we will review the Knowledge Management activities undertaken by the IMI and how they are delivering new services and capabilities that can enhance pharmaceutical R&D.

Sponsored by CERTARA

3:50 Bringing Scientific Data to Life: Agile Data Access and Analysis from Discovery to Development
Jonathan Feldmann, Vice President, Scientific Informatics, Certara
Life sciences research is consistently generating more data, of more complexity, stored in more sources. Use of analytics to derive meaning and make decisions in research and development is hampered by limited access to the data, lack of data integration, and the frequent need for specialist IT resources to support scientists needs. Certara has developed D360 to allow tailored informatics solutions to be rapidly deployed by configuring a standard product to support a wide range of scientific workflows. We will present how organizations across the industry are providing scientists self-service access to dynamically integrated data to make better decisions and accelerate projects, and how these solutions are supporting growing collaborations within global project teams and across organizations.

4:20 Valentine's Day Celebration in the Exhibit Hall with Poster Viewing

5:20 Breakout Discussions in the Exhibit Hall *(see website for details)*

6:30 Close of Day

WEDNESDAY, FEBRUARY 12

7:00 am Breakfast Presentation *(Sponsorship Opportunity Available)* **or Morning Coffee**

» **8:00 PLENARY KEYNOTE SESSION** *(please see page 4 for details)*

9:45 Refreshment Break and Poster Competition Winner Announced in the Exhibit Hall

INTEGRATED R&D INFORMATICS & KNOWLEDGE MANAGEMENT
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INTEGRATED R&D INFORMATICS & KNOWLEDGE MANAGEMENT

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DATA INTEGRATION & SHARING: TOOLS AND POLICIES FOR VISUALIZATION AND ANALYSIS

10:35 Chairperson's Remarks

Larry Hunter, Ph.D., Director, Center for Computational Pharmacology & Computational Bioscience Program, Professor, Pharmacology, University of Colorado

10:40 Knowledge-Based Analysis at Genomic Scale

Larry Hunter, Ph.D., Director, Center for Computational Pharmacology & Computational Bioscience Program, Professor, Pharmacology, University of Colorado

High-throughput instruments and the explosion of new results in the scientific literature is both a blessing and a curse to the bench researcher. Effective design and implementation of computational tools that genuinely facilitate the generation of novel and significant scientific insights remains poorly understood. This talk presents efforts that combine natural language processing for information extraction, graphical network models for semantic data integration, and some novel user interface approaches into a system that has facilitated several significant discoveries.

11:10 Combining Visual Analytics and Parallel Computing for Data-Driven Analysis Pipeline Selection and Optimization to Support the Big Data to Knowledge Transformation

Richard Scheuermann, Director, Informatics, J. Craig Venter Institute

This presentation describes our efforts at the J. Craig Venter Institute (JCVI) in collaboration with the Texas Advanced Computing Center (TACC) in the development of a high performance cyber-infrastructure that combines visual analytics and parallel computing for data-driven selection and optimization of analytical pipelines based on objective performance metrics. We demonstrate the application of these principles and infrastructure for the analysis of genome-wide gene expression and high-throughput, high dimensional flow cytometry data in clinical and translational research settings.

11:40 Co-Presentation: Spotfire Templates for Analysis & Visualization of Project Data

Sandhya Sreepathy, PMP, Head of Operations, Global Discovery Chemistry, Novartis

Heather Hogg, Ph.D., Business Analyst, Novartis Institutes for Biomedical Research

There has been a significant increase in the usage of Spotfire by project teams in Emeryville for visualization and analysis of data. To help streamline development activity and support the needs of project teams with data analysis and visualization, NIBR-IT in collaboration with computational chemistry group in Emeryville developed project based Spotfire templates. Templates were built leveraging existing technologies for retrieval/import of data and incorporated common elements of analysis and visualization(scaffold assignment, ligand, lipophilic efficiency, activity ratios, r-group decomposition etc). Predefined visualizations and filters helped accelerate project team decision making.

12:10 pm Session Break

12:20 Luncheon Presentation *(Sponsorship Opportunity Available)* **or Lunch on Your Own**

1:00 Refreshment Break in the Exhibit Hall and Last Chance for Poster Viewing

HOW BIG DATA WILL DRIVE RESEARCH FORWARD

1:40 Chairperson's Remarks

Michael H. Elliott, CEO, Atrium Research & Consulting LLC

1:45 Harnessing Big Data to Accelerate Drug Development

Vinod Kumar, Ph.D., Senior Investigator, Computational Biology, GlaxoSmithKline Pharmaceuticals

With the rapid development of high-throughput technologies and ever-increasing accumulation of whole genome-level datasets, an increasing number of diseases and drugs can be comprehensively characterized by the changes they induce in gene expression, protein, metabolites and phenotypes. Integrating and querying such large volumes of data, often spanning domains and residing in diverse sources, constitutes a significant obstacle. This talk presents two distinct approaches that utilize these data types to systematically evaluate and suggest new disease indications for new and existing drugs.

2:15 Drug Process Design Improvement based on Data Management and Analysis

Valérie Vermylen, Knowledge Management, Director, GPS, UCB

Most of the scientific process data generated are not free to access, even if managed in databases. At UCB, data was recently made available including its context. It allows process developers to draw easily designed space and define critical parameters. To support investigation studies as impact analysis, manufacturing dashboards and trends are automatically published. An example of correlation between process data and patients' clinical responses will be presented as an illustration of advanced data analysis.

2:45 The Library of Integrated Network-based Cellular Signatures (LINCS) Information FramEwork (LIFE)

Stephan C. Schürer, Ph.D., Associate Professor, Pharmacology, Center for Computational Science at Miller School of Medicine, University of Miami

The NIH-funded LINCS consortium is producing an extensive dataset of cellular response signatures to a variety of small molecule and genetic perturbations. We have been developing the LINCS Information FramEwork (LIFE) – a specialized knowledge-driven search system for LINCS data.

3:15 An Enhanced Molecular Design Platform That Fosters Ideation, Knowledge Transfer, and Collaboration

Sponsored by SCHRÖDINGER

Leah Frye, Ph.D., Vice President, Drug Discovery Applications Group, Schrödinger

Drug discovery is the ultimate team sport. Schrödinger is developing a collaborative and knowledge engineered platform—LiveDesign—to help scientists not only capture their ideas and best practices, but to exploit and share these with select team members. Above and beyond the aggregation of 2D data, this platform will allow users to bring together 3D data with its associated annotations. LiveDesign will ultimately lead to better patient outcomes, promoting better scientific communication by exposing data, ideas, and colleague feedback during the design and redesign phases of molecular discovery.

3:45 Refreshment Break

FROM BIG DATA TO TRANSLATIONAL INFORMATICS

4:00 Chairperson's Remarks

Shoibal Datta, Ph.D., Associate Director, R&D Information Technology, Biogen Idec

4:05 Designing and Building a Data Sciences Capability to Support R&D and Corporate Big Data Needs

Shoibal Datta, Ph.D., Director, Data Sciences, Biogen Idec

To achieve Biogen Idec's strategic goals, we have built a cross-disciplinary team to focus on key areas of interest and the required capabilities. To provide a reusable set of IT services we have broken down our platform to focus on the Ingestion, Digestion, Extraction and Analysis of data. In this presentation, we will outline how we brought focus and prioritization to our data sciences needs, our data sciences architecture, lessons learned and our future direction.

4:35 Translational Informatics: Decomposing to Singularity

John Shon, M.D., Head, Translational Informatics IT, Johnson & Johnson

There has been an explosion of data across discovery, development, and beyond and all informatics groups are struggling with major challenges in computation, storage and analysis. In a large pharmaceutical environment, the value propositions of informatics lie primarily in three dimensions which I describe. In the larger hyperdynamic environments of research technologies, information technologies, and modern science, interdisciplinary and collaborative approaches become imperative to execute translational strategies effectively.

5:05 Integrating Translational Research Tools

Erik Bierwagen, Ph.D., Principal Programmer Analyst, Department of Bioinformatics, Genentech, Inc.

This talk will cover our efforts at creating an integrated informatics system for animal studies from birth to death and beyond. Our efforts span many different disciplines and groups, but share the common effort of integrating data seamlessly.

5:35 Close of Conference Program

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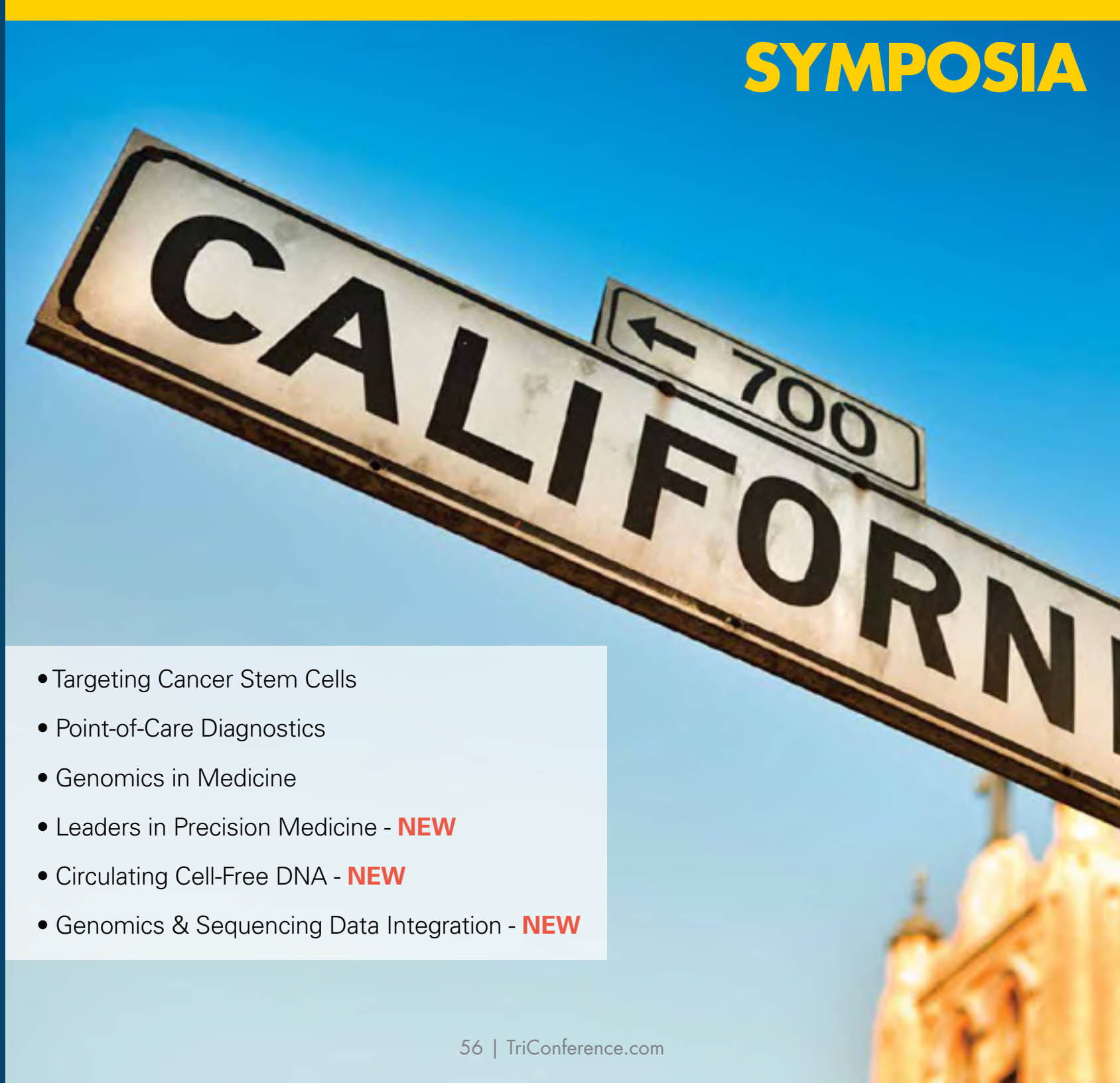

- Targeting Cancer Stem Cells
- Point-of-Care Diagnostics
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Cambridge Healthtech Institute's Third Annual

TARGETING CANCER STEM CELLS

New Opportunities for Oncology Therapeutics

THURSDAY, FEBRUARY 13

7:30 am Registration and Morning Coffee

UNDERSTANDING CANCER STEM CELLS

9:00 Chairperson's Opening Remarks

9:05 Poised with Purpose: Understanding Cell Plasticity in Cancer

Christine Chaffer, Ph.D., Post-Doctoral Fellow, Robert Weinberg Laboratory, Whitehead Institute for Biomedical Research

Here we demonstrate that basal breast cancer non-CSCs are plastic cell populations that readily switch from a non-CSC-to-CSC state. We identify that maintenance of the ZEB1 promoter in a bivalent chromatin configuration enables these cells to switch to a CSC state. Our findings support a dynamic model where interconversions between a low and high tumorigenic state are common occurrences. Inhibiting non-CSC-to-CSC plasticity should be considered as an important adjuvant for current therapeutic regimens.

9:35 Myeloproliferative Neoplasia Remodels the Endosteal Bone Marrow Niche into a Self-Reinforcing Leukemic Niche

Eric Pietras, Ph.D., Passegue Laboratory, University of California, San Francisco

The bone marrow (BM) niche contains specialized cell types necessary for hematopoietic stem cell (HSC) maintenance. We show that secreted and cell-bound factors overproduced by myeloid cells during myeloproliferative neoplasia (MPN) remodels the BM niche into a fibrotic, self-reinforcing leukemic niche that impairs normal hematopoiesis and favors leukemic stem cell (LSC) function. Targeting this pathological interplay could represent a novel avenue for treatment of MPN-affected patients and prevention of myelofibrosis.

10:05 Progression after Chemotherapy and Cancer Stem Cells

Carlos Cordon-Cardo, M.D., Ph.D., Professor, Pathology, Icahn School of Medicine at Mount Sinai

The CSC model comprises an attractive framework to explain acquired chemotherapy resistance, since chemotherapy resistant CSCs would be expected to be well suited to initiate progressive disease. A recent and growing body of evidence supports the contribution of CSCs to chemotherapy resistance across a range of hematological and solid malignancies. For example, CSCs contribute to chemotherapy resistance in prostate cancer, and targeting prostate CSCs has stimulated therapeutic combination strategies that suppress acquired resistance.

10:35 Coffee Break with Exhibit and Poster Viewing

HIGH-THROUGHPUT SCREENING FOR CSC TARGETS

11:05 Novel Methods for Isolating and *in vitro* Expanding CSC for HTS Drug Discovery

Steve McClellan, Manager, Research Operations; Chief, Flow Cytometry & Imaging Core Labs; Coordinator, CSC Working Group, Basic & Translational Sciences, Mitchell Cancer Institute

Traditional methods of isolating CSC from fresh tumors have been difficult. Our laboratory has developed new methods to identify CSC that work across most all tumor types, such as the expression of alkaline phosphatase, and live cell mRNA expression of stem cell markers Oct4, Sox2 and Nanog. We will also present data on the development of new techniques to expand CSC in culture, in such quantities that reliable and reproducible HTS using CSC will now be a reality.

11:35 Beyond WNT, Notch and sHH: Novel CSC Targets

Elaine Hurt, Ph.D., Scientist II, MedImmune

CSCs have high expression of several polycomb repressor members, including EZH2, which is thought to hold the cell in an undifferentiated state. We extended these observations into development of a high throughput assay that obviates the need to isolate CSCs. Using our EZH2 assay, we have identified several novel candidate CSC drug targets in two independent target identification campaigns. This talk will briefly describe this assay and focus on its use in identification of both previously known and unique CSC targets.

12:05 pm Sponsored Presentations *(Opportunities Available)*

12:35 Luncheon Presentation *(Sponsorship Opportunity Available)* **or Lunch on Your Own**

1:05 Session Break

PRECLINICAL ADVANCES TARGETING CSCs

1:50 Chairperson's Remarks

1:55 Development of New Therapeutic Agents that Reduce Tumor Initiating Cell Frequency

Tim Hoey, Ph.D., Senior Vice President, Cancer Biology, OncoMed Pharmaceuticals, Inc.

Cancer stem cells (or tumor initiating cells) mediate tumor progression, metastasis, and recurrence after therapy. We have developed new biologic agents that block key CSC pathways including Notch, Wnt and RSPO-LGR. Currently, we have five therapeutics in clinical testing, anti-DLL4 (demcizumab), anti-Notch2/3, anti-Notch1, anti-FZD (vantictumab), and Fzd8-Fc and others in preclinical development. These agents inhibit tumor growth through multiple mechanisms including a reduction of CSC frequency.

2:25 Targeting Ubiquitination Pathways for Cancer Therapy

Hui-Kuan Lin, Ph.D., Associate Professor, Department of Molecular and Cellular Oncology, The University of Texas MD Anderson Cancer Center

In this talk, I will discuss the role of distinct ubiquitin ligases (E3) in orchestrating non-proteolytic functions in kinase signaling activation. I will also discuss the potential mechanism by which E3 ligases regulate kinase signaling and their role in cancer development. Finally, I will present the evidence that targeting ubiquitination pathways represents a promising therapeutic strategy for cancer stem cells and cancer treatment by using genetic and pharmacological approaches.

2:55 Refreshment Break with Exhibit and Poster Viewing

TARGETING CANCER STEM CELLS
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TARGETING CANCER STEM CELLS

continued

3:25 BNC101: A Novel Therapeutic Antibody Against LGR5 that Inhibits Tumor Growth and Reduces Frequency of Cancer-Initiating Cells

Christopher L. Reyes, Vice President, Research and Development Biologics, Bionomics

BNC101 is a humanized monoclonal antibody that targets LGR5, a 7-transmembrane receptor that is highly specific to cancer stem cells. In multiple preclinical studies, BNC101 blocks the growth of cancer stem cells derived from primary patient tumors both *in vitro* and *in vivo*. Our therapeutic hypothesis is that targeting of CSCs with BNC101 will lead to durable cures in the clinic by eliminating cancer stem cells at the root of cancer.

3:55 Targeting Cancer Stem Cells with 4SC-202 – An Epigenetic Wnt and Hh Inhibitor

Daniel Vitt, Ph.D., CSO, 4SC AG

4SC-202 is a new epigenetic modulator of the Wnt and Hedgehog signaling pathway currently tested in a phase I clinical trial. Comparison with other class I specific HDAC inhibitors shows huge differences in gene regulation and mode of action. By inhibition of the Wnt and Hedgehog signaling pathway, 4SC-202 provokes the inhibition of stemness-related properties. Preliminary data from phase I 'TOPAS' trial and translation of preclinical biomarkers to patient samples will be presented.

4:25 Breakout Discussions *(see website for details)*

5:25 Close of Day

FRIDAY, FEBRUARY 14

8:00 am Morning Coffee

TRANSLATIONAL CONSIDERATIONS

8:25 Chairperson's Remarks

8:30 Strategies to Develop Agents that Preferentially Target Cancer Stem Cells: Clinical Candidates Targeting FAK and PI3K/mTOR

Jonathan A. Pachter, Ph.D., Vice President & Head of Research, Verastem, Inc.

9:00 Antibody-Based Approaches to Targeting Proliferating and Quiescent Cancer Stem Cells

Robert J. Tressler, Ph.D., Vice President, Research & Development, Cellerant Therapeutics, Inc.

9:30 Challenges in Designing Clinical Trials with Stem Cell-Directed Therapies

Anne F. Schott, M.D., Associate Professor, Department of Internal Medicine, University of Michigan

Laboratory research has led to the identification of clinical compounds that target the cancer stem cell population. Clinical trials are being developed to evaluate these stem cell directed therapies, often in combination with a standard treatment such as chemotherapy. The combination therapy paradigm exposes the challenge of demonstrating efficacy and specificity of stem cell targeted agents. New clinical/translational trial endpoints and assays will be necessary to advance the field.

10:00 FEATURED ORAL POSTER PRESENTATION: How to Kill Cancer Stem Cells Without Killing Normal Stem Cells in Adult Patients

William G. Thilly, Sc.D., Professor of Genetics, Toxicology and Biological Engineering, Department of Biological Engineering, Massachusetts Institute of Technology

Through sponsored research program at MIT a series of drugs were identified that specifically kill cancer stem cells but not non-stem cells in cell cultures derived from human pancreatic and colorectal tumors. Several of these drugs are slated for clinical experiments in which pancreatic cancer patients will be treated before surgery to remove their tumors. Using proprietary means the tumors will be dissected and the extent of cancer stem cell killing determined. Those drugs found to be lethal to pancreatic stem cells in patients will be used together in a proprietary regimen one facet of which is designed to overcome the expected resistance of mutant cancer stem cells to individual drugs.

10:30 Coffee Break with Exhibit and Poster Viewing

11:00 Cancer Stem Cell Strategies for Patient Specific Cancer Immunotherapy: Stopping Cancer in Its Tracks

Andrew Cornforth, Ph.D., Cancer Stem Cell Program Manager, California Stem Cell, Inc.

An ideal source of antigens to stimulate an immune response in an immunotherapeutic approach is to use the patient's own tumor. Limitations to this approach are the acquisition of tumor samples in adequate quantities and the lack of antigen sources from the most aggressive phenotypes, namely tumor stem cells. Our approach capitalizes on the recent developments in specific media formulations to isolate and propagate putative cancer stem cells from patient tumor samples to quantities necessary for loading dendritic cells.

CLINICAL UPDATES

11:30 Clinical and Preclinical Studies of Stemline Therapeutics' Clinically Active Agents, SL-401 and SL-701, Directed at Cancer Stem Cells and Tumor Bulk of Hematologic and Brain Cancers

Eric K. Rowinsky, M.D., Head, Research & Development and CMO, Stemline Therapeutics, Inc.

Among its portfolio candidates, Stemline Therapeutics, Inc. is developing two clinical-stage candidates. SL-401 is a targeted therapy directed to the interleukin-3 receptor, which is overexpressed on the CSCs and tumor bulk of many hematologic cancers, whereas SL-701 is a cancer vaccine comprised of synthetic peptides, designed for immune system targeting of both CSCs and TB of brain cancers. These agents have produced regressions in advanced hematologic and brain malignancies, including complete responses and strong survival signals, in phase I/2. Pivotal trials with SL-401 will be initiated in AML and blastic plasmacytoid dendritic cell malignancy, a rare aggressive cancer and unmet medical need, whereas phase 2 trials evaluate SL-701 in children and adults with high-grade glioma.

12:00 pm Dendritic Cell Vaccines Targeting Cancer Stem Cells

John S. Yu, M.D., Professor & Vice Chairman, Neurosurgery, Cedars-Sinai Medical Center; Chairman & CSO, ImmunoCellular

We have developed dendritic cell vaccines for brain cancer. Lead candidate ICT-107 is currently being evaluated in a Phase II, randomized clinical trial. We will present an update on this trial, as well as clinical and immunologic data on the CD133 targeting DC vaccine (ICT-121). A DC vaccine for ovarian cancer has undergone FDA acceptance of an IND and development of a multi-institutional Phase I/II trial will be discussed.

12:30 Close of Symposium

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POINT-OF-CARE DIAGNOSTICS

Enabling Personalized Healthcare Through Rapid Diagnostics

THURSDAY, FEBRUARY 13

7:30 am Registration and Morning Coffee

POINT-OF-CARE ADOPTION: WHAT WILL IT TAKE?

9:00 Chairperson's Opening Remarks
Katherine Tynan, Ph.D., President, Tynan Consulting

9:05 Establish Partnership between Laboratory and Healthcare Providers to Improve Clinical Utility of Point-of-Care Testing
Elsie Yu, Ph.D., DABCC, FACB, Director, Toxicology and Point-of-Care Testing; Associate Director, Chemistry, Department of Laboratory Medicine, Geisinger Health System
Although the cost of point-of-care testing is usually higher than in-lab tests, its ability to give immediate test results makes it appealing to many healthcare providers. Close partnership between laboratory and healthcare providers can ensure proper utilization of point-of-care testing and improve patient care. One must also take into consideration regulatory requirements, operational challenges and implementation strategy to make the best out of these bedside testing devices.

9:35 Point-of-Care Testing Device Design: Practical Tips from the User Perspective
Valerie Ng, M.D., Ph.D., Chair, Laboratory Medicine & Pathology, Alameda Heath System/Highland General Hospital
This presentation will discuss known user point-of-care test performance errors and provide suggestions from the user perspective for device design. Relevant regulatory concerns will be included.

10:05 Implementation of Point-of-Care Testing Programs in Community Pharmacies
Donald Klepser, Ph.D., MBA, Assistant Professor, Pharmacy, University of Nebraska Medical Center College of Pharmacy
As economic pressures mount in healthcare, pharmacists are being placed on the frontline. Development of point-of-care diagnostic tests enable pharmacists to assist with patient management and provide cost-effective care. Health policy makers, pharmacy organizations, and governmental agencies have recognized the potential value of placing point-of-care diagnostic in community pharmacies. This presentation will discuss practical aspects of implementing point-of-care testing in pharmacies.

10:35 Coffee Break with Exhibit and Poster Viewing

THE ROLE OF POC IN COMPANION DIAGNOSTICS

11:05 Why Point-of-Care Technology is Vital for Companion Diagnostics
Peter Miller, COO, Genomic Healthcare Strategies
This presentation will discuss the compelling business reasons for pharma to embrace POC. Selling diagnostics, especially those linked to their drugs, is a new challenge for pharma which has not been fully explored. The old models of pharma selling drugs and reference labs selling tests will change. POC technology will be a critical part of pharma business strategy and offers the opportunity for pharma to better manage the use and profitability of its drugs.

11:35 Use of POC in Oncology Outpatient Practices
Richard A. White, Ph.D., Director, Global Strategy, Breast and Colon Products, Genomic Health, Inc.
With the increased cost of treatment of patients in inpatient settings, economic pressure mounts to reduce overall cost of hospitalization for both acute and chronic treatment. Advances in treatment and diagnostic options have uncovered an opportunity for near-patient testing platforms to serve diagnostic and monitoring purposes.

12:05 pm Sponsored Presentations *(Opportunities Available)*

12:35 Luncheon Presentation *(Sponsorship Opportunity Available)* **or Lunch on Your Own**

1:05 Session Break

STRATEGIES FOR COMMERCIALIZING NEW TECHNOLOGIES

1:50 Chairperson's Remarks
Keith Batchelder, M.D., Founder & CEO, Genomic Healthcare Strategies

1:55 The Role of Evidence-Based Medicine in the Adoption of New POC Technologies
Katherine Tynan, Ph.D., President, Tynan Consulting
Realizing the potential of rapid POC devices to transform the practice of medicine and thereby improve patient outcomes and healthcare cost-effectiveness will be challenging due to misaligned stakeholder interests and the difficulty of changing established medical practice and business models. These barriers, as well as the different stakeholder perspectives, the levels of evidence that can be generated for rapid POC devices and how they can drive change in the healthcare system will be discussed in this presentation.

2:25 Identifying Competitive Advantages for Novel Platform Technologies in Clinical Markets
Rahul Dhanda, Vice President, Marketing, T2 Biosystems
Commercializing new diagnostic platforms requires a clear understanding of the clinical and economic value delivered by the technology, while mapping those advantages to a specific and valuable set of diseases and markets. T2 Biosystems' T2MR technology provides highly sensitive and rapid results in unprocessed samples and up to 25 times faster than gold standard methods. These advances can be applied to multiple diagnostic markets, including sepsis and hemostasis, which provide a case study for commercializing novel platforms.

2:55 Refreshment Break with Exhibit and Poster Viewing

3:25 PANEL DISCUSSION: Making Your Offering and Getting to Market
Moderator: Keith Batchelder, M.D., Founder & CEO, Genomic Healthcare Strategies
Panelists: Robert Di Tullio, Vice President, Global Regulatory and Clinical Affairs, Alere
Rahul Dhanda, Vice President, Marketing, T2 Biosystems
Sailesh Chutani, Ph.D., CEO, Mobisante
Matthew Diamond, M.D., Ph.D., Medical Lead, Misfit Wearables

4:25 Breakout Discussions *(see website for details)*

5:25 Close of Day

POINT-OF-CARE DIAGNOSTICS
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POINT-OF-CARE DIAGNOSTICS

continued

FRIDAY, FEBRUARY 14

8:00 am Morning Coffee

» KEYNOTE SESSION

8:25 Chairperson's Remarks
Peter Miller, COO, Genomic Healthcare Strategies

8:30 Strategic Growth Initiatives, Alliances, and Innovation in Molecular Diagnostics at the Point-of-Care
Michael Nohaile, Ph.D., Vice President, Corporate Strategy, Amgen, Inc.

9:00 Building an Innovative Team: The Key to Success
Craig Lehmann, Ph.D., CC (NRCC), FACB, Dean, The School of Health Technology and Management; Professor, Clinical Laboratory Sciences; Director, Center of Public Health Education, School of Health Technology Management, Stony Brook University
Under the spirit of innovation this presentation is to address opportunities of the many needs of the world's priorities to deliver high impact interventions that will improve health outcomes for their citizens while strengthening health systems.

POC IN GLOBAL HEALTH

9:30 The Changing Landscape in Point-of-Care Diagnostics: Advances in Developing World Applications and Their Potential Impact in Developed Markets
Rich Thayer, Managing Partner, Halteres Associates, LLC; CEO, Catalysis Foundation for Health
Understanding unmet needs and business model options prior to initiating development of POC systems for limited resource settings is essential for success. Learn how advancements in diagnostics technologies and communications, in addition to a better understanding of local market conditions and needs, are driving increased interest in delivering diagnostic solutions for these important markets.

10:00 Microfluidics for Mobile Diagnostics
Samuel K. Sia, Associate Professor, Biomedical Engineering, Columbia University
Lab-on-a-chip (LOC) devices have a tremendous potential for revolutionizing personal health. In developing countries, mobile diagnostics provides immediate diagnosis in the field, and in the U.S., patients and consumers can have greater access to traditionally complex diagnostics. We will discuss our lab's current efforts in these areas, in conjunction with partners in industry, public health, and local governments. Our tests span a variety of technologies, and target HIV, sexually-transmitted diseases, and chronic diseases.

10:30 Coffee Break with Exhibit and Poster Viewing

EMERGING POC AND MOBILE HEALTH TECHNOLOGIES

10:55 Chairperson's Remarks
Michael Nohaile, Ph.D., Vice President, Corporate Strategy, Amgen, Inc.

11:00 The HemoLink: Enabling POC Blood Sampling, Stabilization, Preparation, and Remote Analysis
Erwin Berthier, President, Tasso, Inc.
HIV viral load monitoring is a critical test for HIV management that requires large volumes of blood (>100 uL) and forpatients to locate to an appropriately equipped laboratory or clinic. Tasso has developed the HemoLink to connect patients at home by allowing self-administered blood sampling with the laboratory for remote analysis. The next-generation platform integrates novel technology enabling on-site RNA purification - a step towards POC viral load monitoring.

11:30 Transforming Alzheimer's Diagnosis
Elli Kaplan, CEO, Neurotrack
Neurotrack's technology can predict the onset of Alzheimer's disease three to six years before symptoms occur. Based on groundbreaking research, Neurotrack's technology will enable pharmaceutical companies and CROs to recruit optimal candidates for clinical trials and more effectively measure drug efficacy, speeding up drug development.

11:45 Kinsa Smart Thermometer: Real-Time Health Mapping
Inder Singh, Founder & CEO, Kinsa Health
Kinsa is creating a real-time map of human health, beginning with contagious illness. Using data from mobile-enabled health products, this map will give parents better information as well as help physicians better diagnose and care for patients. The Kinsa Smart Thermometer is a mobile-connected thermometer that allows us to communicate with someone who has just fallen ill, give them the information to get better faster, and collect the data we need to map human health.

12:00 pm Handheld Smartphone Biosensing
Kenny Long, University of Urbana-Champaign
We present a smartphone system based on a custom optical cradle as a spectrometric instrument capable of detecting clinically relevant concentrations of biological analytes using different assay techniques. By translating existing diagnostic tests to a portable system, we open the doors for new applications of proven technologies.

12:15 Ultrasensitive POCT on a Nonfouling Polymer Brush
Ashutosh Chilkoti, Ph.D., Theo Pilkington Chair in Biomedical Engineering; Director, Center for Biologically Inspired Materials and Materials Systems, Duke University
We have developed a POCT in which all reagents are inkjet printed on a polymer brush and which relies upon passive, 2-D diffusion across the brush to generate a colorimetric signal. This assay works with all sandwich immunoassay formats, is quantitative, multiplexable, with sub-picomolar limit-of-detection.

12:30 Close of Symposium

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Cambridge Healthtech Institute's Second Annual

GENOMICS IN MEDICINE

Establishing a Patient-Centric View of Genomic Data

THURSDAY, FEBRUARY 13

7:30 am Registration and Morning Coffee

RETURNING GENOMIC INFORMATION TO THE PATIENT

9:00 Chairperson's Opening Remarks
Jonathan Hirsch, Founder & President, Syapse

» 9:05 KEYNOTE PRESENTATION:

Incidental Findings in Genomic Medicine: The Debate and the Data
Robert C. Green, M.D., MPH, Director, G2P Research Program; Associate Director, Research, Partners Center for Personalized Genetic Medicine, Division of Genetics, Department of Medicine, Brigham and Women's Hospital and Harvard Medical School

Genomics is being rapidly integrated into medicine with many unanswered questions about how and how much risk information should be communicated, and how such information will influence physician and patient behaviors, health outcomes and health care costs. This presentation will summarize data from over 10 years of experimental work in translational genomics and health outcomes, discuss recent ACMG recommendations for incidental findings and preview results from our newest NIH-funded studies, the ongoing MedSeq Project and the recently funded BabySeq Project.

9:35 Genomic Medicine Implementations for Primary Care
Erwin Bottinger, M.D., The Irene and Dr. Arthur Fishberg Professor of Medicine; Director, The Charles Bronfman Institute for Personalized Medicine, Icahn School of Medicine, Mount Sinai

Increasingly, genomic discoveries provide opportunities to personalize medication use and prediction and prevention of common chronic diseases. However, effective integration of genomic medicine in busy primary care practices is hampered by multiple barriers, including provider education gaps and negative impact on clinical workflow. Innovative programs for real-time, point-of-care integration of genomic medicine for primary care providers through genome-informed clinical decision support enabled in electronic health records will be presented.

10:05 Ethical Issues Related to the Return of Incidental Findings in Children/Families
Ingrid A. Holm, M.D., MPH, Director, Phenotyping Core, Program in Genomics, Divisions of Genetics and Endocrinology, Boston Children's Hospital

10:35 Coffee Break with Exhibit and Poster Viewing

EMERGING TOOLS TO ENABLE PHYSICIAN USE

11:05 Reducing the Complexity of Clinical Omics Reporting for Clinicians and Laboratories
Jonathan Hirsch, Founder & President, Syapse

Syapse has built a cloud-based software platform that enables the use of omics at the point of care through an interactive web portal. We will describe how clinical omics labs use the Syapse platform to maintain an evolving omics knowledgebase which drives updated clinical reporting through interactive, intuitive interfaces designed for ease of use and comprehension. We will describe how hospitals use the Syapse platform to place omics results in the context of clinical guidelines, enabling physicians to easily adopt and integrate omics into their clinical workflow.

11:35 Beyond Sequence: Integration of Full-Genome Technologies for Personalized Medicine in the Clinic
Raphael Lehrer, Founder and Chief Scientist, GeneKey

Here we describe how we have used a combination of multiple full genome technologies to triangulate on key dysregulated mechanisms in a patient's sample. By using a combination of systems biology and statistical analysis, we are able to draw conclusions far more precise than one could from sequence alone. We describe how we have applied in the clinic with patients and their oncologists and what we have seen/learned to date, including cases where the dysfunction is not mutation-based.

12:05 pm Targeted NGS of Clinical Samples: Overcoming the Challenges of Obtaining High Quality Data from Low Quality DNA
Sponsored by AsuraGen
Diane Ilsley, Ph.D., Marketing Manager, Genomic Services, Asuragen

12:35 Luncheon Presentation *(Sponsorship Opportunity Available)* **or Lunch on Your Own**

1:05 Session Break

BRIDGING THE GAP BETWEEN RESEARCH AND TREATMENT

1:50 Chairperson's Remarks
Marcia K. Horn, Esq., President and CEO, ICAN

1:55 Genome Sequencing in the Clinic: Found the Variants - Now What?
Jennifer Friedman, M.D., Associate Clinical Professor, Neurosciences and Pediatrics, UCSD/Rady Children's Hospital San Diego

Advances in genome sequencing hold tremendous promise for providing answers and tailored therapies for undiagnosed patients. How to interpret, transmit and act upon volumes of complex data remains a challenge for sequencing providers, physicians and their patients. This presentation will use case-based examples to demonstrate promises and pitfalls encounter along the way.

2:25 The Answer is There but I Don't Understand It: Solutions from the Front Line
Vanya Gant, Ph.D., FRCP, FRCPath, Divisional Clinical Director for Infection, The Department of Microbiology, UCLH NHS Foundation Trust

This talk will introduce the concept and fundamental problem of how to present complex NGS datasets to clinicians – and how this will be critical for rapid uptake. A case study outlining the principles behind a very new and innovative pathology project and way of delivering healthcare diagnostics will also be presented.

2:55 Refreshment Break with Exhibit and Poster Viewing

GENOMICS IN MEDICINE
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GENOMICS IN MEDICINE

continued

3:25 Using a Patient's Genetic Information in the Real World

Michael F. Christman, Ph.D., President and CEO, Coriell Institute for Medical Research

When a patient needs a new prescription, it will be necessary for the physician to quickly and securely access his/her genetic data to understand drug efficacy prior to dosing. Who will patients and doctors 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 have proposed solutions that are generally applicable.

3:55 Developing Clinical Sequencing Assays at Einstein-Montefiore

Cristina Montagna, Ph.D., Associate Professor, Genetics, Albert Einstein College of Medicine

We developed a program to introduce Next-Generation Sequencing (NGS) to address the needs of individuals receiving clinical care at Montefiore Medical Center. After extensive dialogue with clinicians, we designed a custom gene panel, spanning 5Mb and consisting of 650 genes targeting known Mendelian loci, some pediatric diseases and several hotspot genes in various cancer types. By building a basic infrastructure for transitioning NSG in the clinic we have encountered roadblocks and established protocols to overcome these.

4:25 Breakout Discussions *(see website for details)*

5:25 Close of Day

FRIDAY, FEBRUARY 14

8:00 am Morning Coffee

THE IMPACT OF DTC TESTING

8:25 Chairperson's Remarks

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

8:30 Direct-to-Consumer Genetic Testing: Balancing the Good and the Bad

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

There have been active debates in the media and the science community on whether the growing trend of consumer directed tests could lead to unintended harm or empower people. Genetic information, whether it is used for prediction of disease risk, therapeutic decisions or diagnosis of disease is empowering. This talk will discuss analytical validity, clinical validity and clinical utility of genetic tests and discuss, using specific examples, which types of DTC genetic tests can be helpful and where they can be misleading.

9:00 Crowdsourcing Genetic Discovery

Nicholas Eriksson, Ph.D., Principal Scientist, Statistical Genetics, 23andMe

As the flurry of genetic progress in the last decade has created an excitement about genetics in the public, 23andMe has brought together hundreds of thousands of consumers to learn about their genetics and to participate in new research online. I'll show examples of how this approach can lead to new discoveries, discuss the current state of risk prediction based on genetics, and give some projections for the future.

9:30 Personal Genomics through Smart Digital Media

Patrick Merel, Ph.D., Founder & CEO, Portable Genomics

Portable Genomics will be sharing the concept of its mobile platform that facilitates the access to important genomic information through the use of familiar interfaces similar to the digital media industry.

10:00 Sponsored Presentations *(Opportunities Available)*

10:30 Coffee Break with Exhibit and Poster Viewing

11:00 The Ethical and Social Implications of Direct-to-Consumer Genetic Testing

Sandra Soo-Jin Lee, Ph.D., Senior Research Fellow, Center for Biomedical Ethics, Stanford University Medical School

This presentation will examine the ethical and social implications of DTC personal genomics. Network ethnography will be used to trace how and with whom individuals share their personal genomic information, to examine attitudes and perspectives among DTC PGI consumers, and to investigate how companies create online tools which form strategic collaborations and networks.

IMPACT AND EVOLVING ROLE OF GENETIC COUNSELING

11:30 Next-Generation Genetic Counseling

Ramji Srinivasan, CEO & Co-Founder, Counsyl

12:00 pm TDTC(CC) – Consumers, Clinicians and Counseling

Erica Ramos, MS, CGC, Clinical Genomics Specialist, Certified Genetic Counselor, Translational and Consumer Genomics, Illumina, Inc.

Direct-to-consumer (DTC) testing has raised various concerns among medical professionals, including issues regarding communication of results and integration of tests into patient care. However, DTC testing is a key driver of patient interest in genomic medicine and provides an opportunity for clinicians and genetic counselors to increase awareness of the benefits of genomics and the services that the genetics community can provide. This talk will focus on the applications and positioning of DTC testing from the genetic counselor's perspective.

12:30 Close of Symposium

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Cambridge Healthtech Institute's Inaugural

LEADERS IN PRECISION MEDICINE

Regulatory, Economic and Data Dilemmas in a Genomic Era

THURSDAY, FEBRUARY 13

7:30 am Registration and Morning Coffee

ECONOMIC AND DEVELOPMENT MODELS

9:00 Chairperson's Opening Remarks

Mark Trusheim, Executive in Residence & Visiting Scientist, MIT; former Special Government Employee, FDA

9:05 Precision Medicine Economics: Breakthrough or Breakdown?

Mark Trusheim, Executive in Residence & Visiting Scientist, MIT; former Special Government Employee, FDA

Precision medicines represent over half of the drugs receiving FDA Breakthrough designation. While patients are poised to receive great benefits, the economics of developing and reimbursing precision medicines may break both innovators and payers. A breakthrough in precision medicine economics that benefits all is possible, if we cleverly combine next-generation science, big data, creative business models and innovative public policies.

9:35 Collaborative Models for Rx/Dx Partnering and Impact in the Immunology Therapeutic Area

Mark Curran, Ph.D., Vice President, Systems Pharmacology & Biomarkers, Immunology Therapeutic Area, Janssen Pharmaceuticals, Inc.

10:05 CASE STUDY 1: A Systems Approach to Pharmaceutical R&D

Robert J. Mulroy, President & CEO, Merrimack Pharmaceuticals

Merrimack is an oncology-focused biotechnology company developing targeted therapies coupled with companion diagnostics. The company is founded on a systems biology approach to R&D that integrates big data biology, computational modeling, and systems engineering to understand the multi-dimensional interactions that regulate cellular networks. The talk will focus on how the organizational and scientific systems approaches employed by Merrimack have impacted productivity of discovery and preclinical research and is seeking to improve the productivity of clinical research to create precision medicines in cancer.

10:35 Coffee Break with Exhibit and Poster Viewing

COMPANION DIAGNOSTICS FOR STRATIFIED MEDICINES

11:05 Demonstrating Clinical Utility & Evidence Generation

Mara Aspinall, President and CEO, Ventana Medical Systems, Inc.

11:25 The Role of LDTs Before and After the Approval of a Companion Diagnostic

Terry Robins, Ph.D., former Global Director, Biomarker R&D, Quest Diagnostics

Laboratory-Developed Tests (LDTs) can play a significant role in the transition from experimental Biomarker assays to Companion Diagnostics. LDTs represent a flexible, cost-effective approach to mitigate the risks involved in developing a successful companion diagnostic strategy. Even after regulatory approval of both a drug and its companion diagnostic, LDTs serve to enhance the companion diagnostic approach and are required to improve and refine patient selection based on the most current scientific evidence.

11:45 Turbulent Change and the Reimbursement of Companion Diagnostics

Bruce Quinn, M.D., Ph.D., Senior Health Policy Specialist, Foley Hoag, LLP

In 2012/2013, CMS announced six policy efforts regarding diagnostic payments: The MOLDX coverage/pricing pilot program, genetic test price cuts, policies for multi-analyte test payments, and three CY2014 proposals: (a) cap the prices of advanced pathology tests like *in situ* hybridization, (b) bundle all diagnostic tests to physician visits or procedures for hospital outpatients, and (c) revise the entire clinical chemistry fee schedule due to technological changes. The outlook for 2014/2015 will be discussed.

12:05 pm Sponsored Presentations *(Opportunities Available)*

12:35 Luncheon Presentation *(Sponsorship Opportunity Available)* **or Lunch on Your Own**

1:05 Session Break

1:50 Chairperson's Remarks

1:55 PANEL DISCUSSION: Precision Medicine Opportunities with Legacy Drugs

Moderator: Bruce Quinn, M.D., Ph.D., Senior Health Policy Specialist, Foley Hoag, LLP

With few exceptions, precision medicine approaches are just being applied to new therapeutics despite our knowledge that current, legacy drugs provide benefits to only about half of the patients taking them. What is preventing us from bringing precision medicine to these therapies? And if we succeed, what would be the benefits to patients, payers and the innovators?

Panelists:

Mark Curran, Ph.D., Vice President, Systems Pharmacology & Biomarkers, Immunology Therapeutic Area Janssen, Pharmaceutical Companies of Johnson & Johnson

James Creeden, M.D., Ph.D., CMO, Roche Diagnostics

Mark Trusheim, Executive in Residence & Visiting Scientist, MIT; former Special Government Employee, FDA

Additional Panelist to be Announced

2:55 Refreshment Break with Exhibit and Poster Viewing

TRANSFORMING THE REGULATORY PARADIGM

3:25 PANEL DISCUSSION: Adaptive Licensing: Evolving Regulation to Enable Precision Medicine

Moderator: Mark Trusheim, Executive in Residence & Visiting Scientist, MIT; former Special Government Employee, FDA

Traditional regulatory decision-making involves the evaluation of safety and efficacy evidence from studies of phenotypically-defined patient populations. Current evidence standards for regulation reflect the predictive failure of these parameters, and other available tools, for more precisely identifying meaningful subpopulations with respect to their potential for benefit/harm. What are the implications of the emerging tools of precision medicine for regulation, and vice versa? How might we think about evidence, regulation, and, more broadly, innovation as we move from population-based drug development to treatment optimization for an N of 1? A multi-stakeholder panel from the MIT New Drug Development Paradigms (NEWDIGS) collaboration will share their perspectives from a series of live simulation exercises that have formed the foundation for a European-wide initiative now being planned for launch in 2014.

Panelists:

Hans-Georg Eichler, M.D., Senior Medical Officer, European Medicines Agency (EMA)

Kenneth Oye, Ph.D., Associate Professor Political Science & Engineering Systems, MIT

Edmond Pezalla, M.D., Ph.D., National Medical Director for Pharmacy Policy and Strategy, Aetna

Thomas Unger, Ph.D., Executive Director, Worldwide Regulatory Strategy, Pfizer, Inc.

4:25 Breakout Discussions

5:25 Close of Day

LEADERS IN PRECISION MEDICINE
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LEADERS IN PRECISION MEDICINE

continued

FRIDAY, FEBRUARY 14

8:00 am Morning Coffee

TRANSLATING SCIENTIFIC POSSIBILITIES INTO PATIENT TREATMENT REALITITES

8:25 Chairperson's Remarks

8:30 CASE STUDY 2: Kalydeco – Targeting the G552D Population in Cystic Fibrosis

Peter Mueller, Ph.D., CSO & Executive Vice President, Global Research & Development, Vertex Pharmaceuticals

To date, more than 1900 different genetic mutations are known in the gene encoding the Cystic Fibrosis Transmembrane Regulator (CFTR) protein. KALYDECO, recently approved by the FDA and EMA, is a breakthrough therapeutic approach to treat the underlying root cause of CF in patients with the G551D mutation by enhancing chloride transport using a CFTR potentiator. Relevant biological concepts and clinical development strategies addressing different aspects of the complex interplay between genotype and phenotype in patients with Cystic Fibrosis will be discussed.

9:00 Demonstrating Clinical Utility/Health Technology Assessment

Naomi Aronson, Ph.D., Executive Director, Technology Evaluation Center, Blue Cross Blue Shield

9:30 Bringing Comprehensive Molecular Information into Routine Clinical Care

Josephine N. Harada, Ph.D., Director, Strategic Alliances, Foundation Medicine, Inc.

10:00 Sponsored Presentations *(Opportunities Available)*

10:30 Coffee Break with Exhibit and Poster Viewing

11:00 The "$1000 Genome": Panacea or Pandora's Box?

Kathryn Phillips, Ph.D., Professor, Health Economics and Health Services Research, University of California, San Francisco

New technologies are enabling the arrival of the much-awaited "$1000 genome"—the ability to sequence an individual's or a tumor's entire genome quickly and relatively inexpensively [whole genome sequencing (WGS)]. WGS is now being offered in clinical care and is expected to become more widely used in the near future. However, this technological advance threatens to outpace our ability to use it effectively in clinical practice and to address the associated health policy issues. This talk will discuss work on evaluating the potential benefit-risk tradeoffs of WGS from the perspectives of patients, providers, the health care delivery system, and society.

11:30 PANEL DISCUSSION: The Promise of Patient Data in Healthcare

The generation and systematic utilization of vast amounts genomic data promises to be the key in enhanced decision-making for the patient care, diagnosis, and the development of targeted treatments. This panel will discuss current challenges in implementing genomic data into modern-day healthcare, such as assessing the emerging role of next-generation sequencing (NGS) in clinical trial design; benefit-risk tradeoffs of NGS, clinical utility and health technology assessment; data privacy and sharing.

Panelists:

Peter Mueller, Ph.D., CSO & Executive Vice President, Global Research & Development, Vertex Pharmaceuticals

Naomi Aronson, Ph.D., Executive Director, Technology Evaluation Center, Blue Cross Blue Shield

Kathryn Phillips, Ph.D., Professor, Health Economics and Health Services Research, University of California, San Francisco

Josephine N. Harada, Ph.D., Director, Strategic Alliances, Foundation Medicine, Inc.

Michael Christman, Ph.D., President and CEO, Coriell Institute for Medical Research

12:30 Close of Symposium

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Cambridge Healthtech Institute's Inaugural

CIRCULATING CELL-FREE DNA

Advancing Noninvasive Diagnostics

THURSDAY, FEBRUARY 13

7:30 am Registration and Morning Coffee

CIRCULATING TUMOR DNA: MONITORING DISEASE AND TREATMENT

9:00 Chairperson's Opening Remarks

Abhijit A. Patel, M.D., Ph.D., Assistant Professor, Department of Therapeutic Radiology, Yale University School of Medicine

» 9:05 KEYNOTE PRESENTATION:

Plasma DNA Sequencing for Noninvasive Prenatal Testing and Cancer Detection

Dennis Lo, M.D., Ph.D., Director, Li Ka Shing Institute of Health Sciences, The Chinese University of Hong Kong

Over the last 5 years, there is much interest in the use of massively parallel sequencing of plasma DNA for molecular diagnostics. Hence, fetal chromosomal aneuploidies, genome-wide molecular karyotyping and even fetal whole genome sequencing have been accomplished through the sequencing of maternal plasma DNA. We have also shown that this approach can be used for the noninvasive detection of tumor-associated copy number aberrations and single nucleotide mutations in cancer patients.

9:35 Detecting Oncogenic Mutations from Circulating Tumor DNA (ctDNA): Lessons Learned from ctDNA Analysis in Clinical Trials

Elizabeth Punnoose, Scientist, Oncology Biomarker Development, Genentech

ctDNA is an attractive alternative to patient tumor biopsies to detect oncogenic mutations. Its value is in complementing tumor-based analysis to determine patients' current mutation status, longitudinal monitoring on treatment, as well as detection of resistance mutations as they arise. Results from ctDNA analysis from clinical trials of targeted agents will be discussed.

10:05 Next-Gen Sequencing to Track Changes in Circulating Tumor DNA

Abhijit A. Patel, M.D., Ph.D., Assistant Professor, Department of Therapeutic Radiology, Yale University School of Medicine

An ultrasensitive, multi-target assay will be presented that can identify and quantify mutant ctDNA using error-suppressed next-generation sequencing. This assay is able to monitor treatment response and disease progression in patients with non-small cell lung cancer, without prior knowledge of the mutation profile of their tumor.

10:35 Coffee Break with Exhibit and Poster Viewing

11:05 Circulating Tumor DNA for Noninvasive Cancer Diagnostics

Dana Tsui, Ph.D., Postdoctoral Research Fellow, Rosenfeld Lab, Cancer Research UK Cambridge Institute, University of Cambridge

This talk will review applications of circulating tumor DNA as a "liquid biopsy" to monitor cancer dynamics. Clinical cases will be presented to demonstrate the potential to monitor cancer patients' response to treatment, and to identify *de novo* genomic changes that are linked to drug resistance. Practical considerations associated with the clinical implementation of circulating tumor DNA analysis will also be discussed.

11:35 Evaluation of EGFR Mutations in Plasma from NSCLC Patients: Utility in Managing Patients on TKI Therapy

Mitch Raponi, Ph.D., Senior Director, Molecular Diagnostics, Clovis Oncology

We are utilizing blood-based molecular testing to determine resistance mutation profiles in these patients with the goal of enabling targeted subsequent therapy without need for repeat lung biopsy. The utility of plasma-based EGFR mutational analysis will be described in the context of CO-1686, a novel third-generation TKI that selectively inhibits the EGFR activating and T790M resistance mutations in NSCLC patients.

12:05 pm Detection and Monitoring of BRAF and KRAS Mutations in Cell-Free Urinary DNA of Metastatic Cancer Patients

Sponsored by TROVAGENE CELL-FREE MOLECULAR DIAGNOSTICS

Mark Erlander, Ph.D., CSO, Trovagene

Detection and monitoring of oncogenic mutations in cell-free urinary DNA opens the possibility of a new paradigm for a truly non-invasive method of individualized care for metastatic cancer patients, which would enable the quantitation of mutational tumor load and respective concordance to therapeutic responsiveness followed by detection of emerging genomic alterations underlying acquired resistance. Next-generation sequencing techniques enable monitoring of key driver mutations and resistance mechanisms using targeted gene panels.

12:35 Luncheon Presentation *(Sponsorship Opportunity Available)* **or Lunch on Your Own**

1:05 Session Break

APPLICATIONS IN NONINVASIVE PRENATAL DIAGNOSTICS

1:50 Chairperson's Remarks

Subhashini Chandrasekharan, Ph.D., Assistant Research Professor, Institute for Genome Sciences & Policy, Duke University

1:55 Fetal Cell-Free DNA: How the Fraction and Amount Informs a Noninvasive Prenatal Test Result

Sponsored by Ariosa DIAGNOSTICS

Arnold Oliphant, Ph.D., CSO, Ariosa Diagnostics

Cell free plasma DNA can be highly variable. Maternal factors, shipping, handling and laboratory processing can all have an influence on the plasma DNA that is evaluated for NIPT testing. It is important to evaluate these effects in each sample to assure that NIPT results provide added value to the patient.

2:10 Commercialization of Noninvasive Prenatal Testing: Ethical and Policy Implications

Subhashini Chandrasekharan, Ph.D., Assistant Research Professor, Institute for Genome Sciences & Policy, Duke University

The introduction of current and emerging noninvasive prenatal testing (NIPT) into clinics in the US and abroad raises both practical and ethical challenges. This talk will briefly discuss ethical, legal, social and practical issues associated with commercialization of NIPT technologies and their policy implications.

2:40 High Risk Screening and Outlook for the Future

Kumar Duraiswamy, M.D., MBA, Associate Director, Field Medical Affairs, Illumina, Inc.

In this talk, we examine the sequencing technologies that provide the framework for noninvasive prenatal testing (NIPT). This presentation will also compare and contrast the commercially available noninvasive prenatal tests in the United States, discusses clinical implementation recommendations from professional societies and highlights considerations for genetic counseling.

3:10 Refreshment Break with Exhibit and Poster Viewing

CIRCULATING CELL-FREE DNA
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CIRCULATING CELL-FREE DNA

continued

3:40 Initial Commercial Results from a Noninvasive Prenatal Aneuploidy Test that Employs Massively Multiplexed Targeted PCR Amplification and Sequencing of 19,488 SNPs

Matthew Hill, Ph.D., Vice President, Research, Natera, Inc.

Targeting tens of thousands of SNPs represents a significant advance over shotgun methods used to noninvasively detect fetal chromosome copy number abnormalities. This versatile platform generates array-like capabilities from PCR and NGS, yielding highly quantitative data to which robust Maximum Likelihood Estimation methods can be applied. We report here very high sensitivity and specificity achieved using this method, and its adaptation for detection of microdeletions in ccfDNA and other mixed samples.

4:10 Noninvasive Personalized Genomics

Charles R. Cantor, Ph.D., CSO, Sequenom

Cell-free DNA in plasma reflects cell death in a variety of host compartments. Differences in sequence, copy number or epigenetics among these compartments can be used to access DNA samples without invasive procedures. Depending on the application the circulating DNA can be analyzed by random shotgun sequencing, targeted sequencing, DNA mass spectrometry or PCR. Some examples of clinical applications of these approaches will be described.

4:40 Breakout Discussions *(see website for details)*

5:25 Close of Day

FRIDAY, FEBRUARY 14

8:00 am Morning Coffee

NEW APPROACHES AND FINDINGS

8:25 Chairperson's Remarks

Mitch Raponi, Ph.D., Senior Director, Molecular Diagnostics, Clovis Oncology

8:30 Rapid Detection of Cancer-Related Circulating Cell-Free (CCF) DNA Directly from CLL Patient Blood

Michael J. Heller, Ph.D., Professor, Bioengineering and Nanoengineering, University of California, San Diego

We now demonstrate a dielectrophoretic (DEP)-based approach that allows ccf-DNA biomarkers from chronic lymphocytic leukemia (CLL) patients to be isolated in less than 10 minutes directly from a small volume (20ul) of unprocessed whole blood. The use of DEP devices for rapid isolation of ccf-DNA directly form a small volume of blood has considerable potential as a noninvasive point-of-care (POC) approach for the detection of incipient, residual, and recurrent cancer.

9:00 Nanotechnology Enhanced Analysis of Methylation and Integrity Index of Circulating Tumor DNA

Jeff Tza-Huei Wang, Professor, Mechanical Engineering & Biomedical Engineering, Sidney Kimmel Comprehensive Cancer Center, Institute for NanoBioTechnology, Johns Hopkins University

This talk describes a streamlined DNA methylation detection platform that utilizes silica super paramagnetic particles to improve the processing of circulating DNA in serum/plasma and quantum dots to enhance methylation detection. In addition, a microfluidic single molecule detection platform for analyzing the integrity index of circulating DNA as a potential cancer marker is also discussed.

9:30 Circulating Cell-Free MicroRNAs as Biomarkers

Muneesh Tewari, M.D., Ph.D., Human Biology, Fred Hutchinson Cancer Research

10:00 Sponsored Presentations *(Opportunities Available)*

10:30 Coffee Break with Exhibit and Poster Viewing

ALTERNATE APPLICATION AREAS FOR CFDNA

11:00 Sequencing of Plasma DNA for the Diagnosis of Infection and Rejection in Organ Transplantation

Iwijn De Vlaminck, Ph.D., Researcher, Quake Lab, Stanford University

In this talk, I will present results of our efforts to develop DNA-sequencing based diagnostics of both rejection and infection in organ transplantation. We have analyzed cell-free DNA in plasma samples from a large cohort of transplant recipients (>700 samples, >100 transplant recipients). We find that donor-derived cell-free DNA is an informative marker of rejection. An analysis of non-human derived sequences reveals strong interactions between the virome and immune strength.

11:30 Cell-Free DNA: A Noninvasive Monitoring Tool for Renal Injury

Minnie Sarwal, M.D., FRCP, DCH, Ph.D., Professor, Pediatrics and Transplant Nephrology, CPMC; Director, The BIOMARC Program for Personalized Medicine, CPMCRI, Sutter Health Care

12:00 pm Prognostic Utility of Cell-Free DNA in Sepsis: Basic and Translational Studies

Dhruva Dwivedi, Ph.D, Research Associate, Department of Medicine, Division of Hematology, David Braley Research Institute, Thrombosis and Atherosclerosis Research Institute (TaARI), McMaster University

Sepsis remains a major cause of morbidity and mortality in ICU patients. Our studies focus on the mechanistic links between cell-free DNA, blood coagulation, and inflammation. My presentation will highlight findings from *in vitro* studies as well as translational studies in septic patients and mouse models of sepsis.

12:30 Close of Symposium

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Cambridge Healthtech Institute's Inaugural

GENOMICS & SEQUENCING DATA INTEGRATION, ANALYSIS AND VISUALIZATION

Converging Cloud Computing and Big Data to Support Life Sciences Research

THURSDAY, FEBRUARY 13

7:30 am Registration and Morning Coffee

GENOME BIG DATA: CHALLENGES AND OPPORTUNITIES

9:00 Chairperson's Opening Remarks
Scott Kahn, Ph.D., CIO and Vice President, Informatics, Illumina

» 9:05 KEYNOTE PRESENTATION:

Genomic Big Data: Benefits and Challenges of Large Scale Information Aggregation
Scott Kahn, Ph.D., CIO and Vice President, Informatics, Illumina
The recent trend towards the large-scale aggregation of genomic and phenotypic information creates several unique benefits to advancing many areas of the life sciences that belies the challenges to the practitioner. Such "genomic Big Data" can be viewed from a variety of overlapping perspectives that all must converge for a practical solution to emerge. This presentation will introduce a framework for dissecting these challenges and will discuss progress to date on achieving practical solutions for the scientist as well as the informatician.

9:35 High-Performance Access to Large, Diverse Genomics Data Set
Carl Meinhof, Ph.D., Manager, Research Informatics, IT, Ceres, Inc.
Scientists expect to navigate genomic data with the same ease and speed that they can navigate geographic data. We have developed a genome browser that uses algorithms from game development to provide high-performance visualization of genomics data. Data from multiple sources can be integrated in a relational database backend, but users can also visualize data from files. The database can be hosted in the cloud to facilitate sharing of data. Due to its high speed and ease of use the browser enables playful exploration of data. This presentation demonstrates live examples of how the application can be used and how it performs.

10:05 Integrative Analyses on Clinical Transcriptomics for Drug Discovery Programs
Deepak K. Rajpal, D.V.M., Ph.D., Director, Computational Biology, GlaxoSmithKline
Integrative analyses offer the power to bring together data from multiple sources. We will present a brief overview of the studies we have conducted for drug discovery programs.

10:35 Coffee Break with Exhibit and Poster Viewing

DATA INTEGRATION AND BUSINESS INTELLIGENCE

11:05 Bioinformatics in the Amazon Cloud
Angel Pizarro, Senior Solutions Architect, Amazon Web Services
Learn how health care and life sciences organizations are leveraging the integration between Amazon DynamoDB, Amazon Elastic MapReduce, and Amazon Redshift to manage and compute their data at high scale for the entire data lifecycle: from creation to analysis. In this session, we will provide an introduction to Amazon Web Services, plus we will describe 21st century architecture design patterns leveraging cloud computing, and finally we will highlight a couple of customer success stories in the biomedical and life sciences industries. Using existing SQL-based tools and business intelligence systems in the Amazon cloud, you will learn how to gain deeper insight from your data at lower cost and without the traditional headaches of managing your own infrastructure.

11:35 Scaling Systems for Research Computing
Adam Kraut, Scientific Consultant, BioTeam

12:05 pm Machine Learning Approaches to Predicting 7-day Hospital Readmission in Children
Saras Saraswathi, Ph.D., Clinical Instructor, Pediatrics, Ohio State University; Postdoctoral Research Associate, Battelle Center for Mathematical Medicine, Research Institute, Nationwide Children's Hospital

12:35 Luncheon Presentation *(Sponsorship Opportunity Available)* **or Lunch on Your Own**

1:05 Session Break

BIOINFORMATICS AND CLOUD COMPUTING TOOLS FOR SCIENCE

1:50 Chairperson's Remarks
Ted Slater, Senior Solutions Architect, Life Sciences, YarcData, a Cray Company

1:55 OpenBel: Data Standards and Knowledge Engineering for the Life Sciences
Ted Slater, Senior Solutions Architect, Life Sciences, YarcData, a Cray Company
The recent emphasis on big data and cloud computing has brought with it a sharper focus on data-centricity and infrastructure convergence. While these are excellent goals in principle, they are very difficult to achieve, in large part because of legacy knowledge representation and architecture choices that work primarily to create data silos. Data silos, in turn, are brittle, non-interoperable solutions that can severely hinder modern data infrastructure efforts. OpenBeL is an open source knowledge representation standard, together with a set of software tools, that can help eliminate data silos and fully enable knowledge-based life sciences research.

2:25 Mining the Human Immune System with NGS, AbGenesis & the Cloud
Giles Day, CEO, Distributed Bio
Using NGS sequencing it is now possible to gain insights into how antibody repertoires respond and adapt during treatments such as vaccination, immunomodulation and tumor suppression. The millions of sequence reads and complexity of the data require the adoption of powerful algorithms which in turn require enormous compute resources. This talk will give examples of how simple tools can now be used by bench scientists to mine the immune system.

2:55 Refreshment Break with Exhibit and Poster Viewing

GENOMICS & SEQUENCING DATA INTEGRATION, ANALYSIS AND VISUALIZATION
CONTINUED ON NEXT PAGE

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GENOMICS & SEQUENCING DATA INTEGRATION, ANALYSIS AND VISUALIZATION

continued

3:25 Integrated Research Data Management and Analysis in NGS Using Globus Genomics
Ravi Madduri, Fellow, Computation Institute, University of Chicago; Project Manager, Math and Computer Science Division, Argonne National Lab
In this talk we will present Globus Genomics. Globus Genomics is a robust, scale on-demand solution that provides end-to-end research data management for Next-Gen Sequencing Analysis using Galaxy, Globus Online and Amazon Web Services. The emphasis is on providing the researcher with a high degree of flexibility to inspect, customize, and configure NGS analysis tools and workflows, and share findings with collaborators.

3:55 A Multi-Center Biomarkers Knowledge Environment for NCI's EDRN Early Detection Cancer Research Program
Daniel Crichton, Informatics PI, NASA's Jet Propulsion Laboratories
NASA Jet Propulsion Laboratory and the National Cancer Institute have developed a comprehensive knowledge environment to support the capture, processing, management, analysis and distribution of results from biomarker research generated from the Early Detection Research Network (EDRN). The knowledge environment leverages a distributed, open source infrastructure, originally developed at NASA's Jet Propulsion Laboratory, to support scientific data management, archiving and distribution for NASA's planetary and Earth robotic missions. The knowledge environment leverages modern informatics technologies for bringing the multi-center EDRN into a distributed, virtual enterprise. This talk will introduce the project, describe the transfer of technologies between space and cancer research, and lessons learned in building a national enterprise.

4:25 Breakout Discussions *(see website for details)*

5:25 Close of Day

FRIDAY, FEBRUARY 14

8:00 am Morning Coffee

DATA SECURITY: STAYING SAFE IN THE CLOUD

8:25 Chairperson's Remarks
Peter Alterman, Ph.D., COO, SAFE-BioPharma Association

8:30 An Expert's Guide through the Identity Landscape
Peter Alterman, Ph.D., COO, SAFE-BioPharma Association

9:00 An Infrastructure Approach for Securing and Scaling Data Collaborations
Michael Shoffner, Senior Research Software Architect, Renaissance Computing Institute (RENCI)/University of North Carolina Chapel Hill (UNC); Adjunct Instructor, School of Information and Library Science (SILS), UNC
RENCI is developing an infrastructure that combines policy based data management, software defined networking, and endpoint security to create a cloud based fabric that enables secure collaborative data management over large scientific and medical data sets. Components of this architecture are already in production use by UNC's Lineberger Comprehensive Cancer Center, SAS, and NC TraCS, the National Institutes of Health (NIH) Clinical Translational Science Award (CTSA) institute at UNC. This talk will outline how this open source architecture works and how to employ it in infrastructure solutions.

9:30 Security and Control in the Cloud
Ramin Daron, IT Director, Information Technology, Johnson & Johnson
Richard Wolf, Senior Director, Global Medical Safety - Pharmacovigilance Operations, Johnson & Johnson
This presentation will cover practical implementations of security and safeguards for a cloud service in a regulated industry. Concepts of data ownership and security, regulations, policies and architectural decisions to be offered for consideration.

10:00 Talk Title to be Announced
Adam Fuchs, CTO and Co-founder, Sqrrl

10:30 Coffee Break with Exhibit and Poster Viewing

FEATURED SESSION

Sponsored by THOMSON REUTERS

10:55 Chairperson's Remarks
Andreas Matern, Vice President, Disruptive Innovation, Life Sciences, Thomson Reuters

11:00 Advances in Translational Approaches in Brain Diseases
Sirimon O'Charoen, Ph.D., Manager, Translational Medicine, Life Sciences Professional Services, Thomson Reuters

11:30 Making Sense of Big Data in Pharma
Andreas Matern, Vice President, Disruptive Innovation, Life Sciences, Thomson Reuters
Big Data is quickly becoming an overused, and poorly understood, term in technology. This talk will focus on Big Data for the life sciences: are -omics data the only 'big data'? What's a practical working definition for Big Data in the Life Sciences and does it differ from other areas where data is analyzed at scale? What role does visualization have in Big Data? How do we resolve gaps in life sciences data? How can we spot trends utilizing aggregated information from disparate data sources, and can we effectively ask questions and monitor the ever growing amount of structured and unstructured content that we have access to?

12:00 pm Finding a Needle in a Haystack – Making Sense of Gene Variant Information
Chris Willis, Ph.D., Solution Specialist, Life Sciences, Thomson Reuters
Rapidly advancing 'Next Gen Sequencing' (NGS) technologies led to generation of massive amounts of sequencing data which contain valuable information about correlation between genome variations and clinical phenotypes. Yet this information remains fragmented, scattered among multiple databases and individual publications. This prevents its effective use for the interpretation of genomic data, which increasingly becomes a bottleneck in diagnostic applications. To address this problem, we have indexed a database of gene variant content which integrates knowledge on tens of thousands of gene variants and their reported implication to health, collected from a broad range of sources. All information is manually curated, and describes the association between a genetic variant and disease or treatment response. Each record provides the level of correlation, effect of the genotype-phenotype association, and possible role as a biomarker, delivering a level of confidence in the variant relationship. Programmatic access to this gene variant content makes this data available for incorporation into internal systems and workflows as well as filtering of those clinically actionable gene variants related to genetic predisposition to disease and sensitivity/resistance as a predictor of response to therapies.

12:30 Close of Symposium

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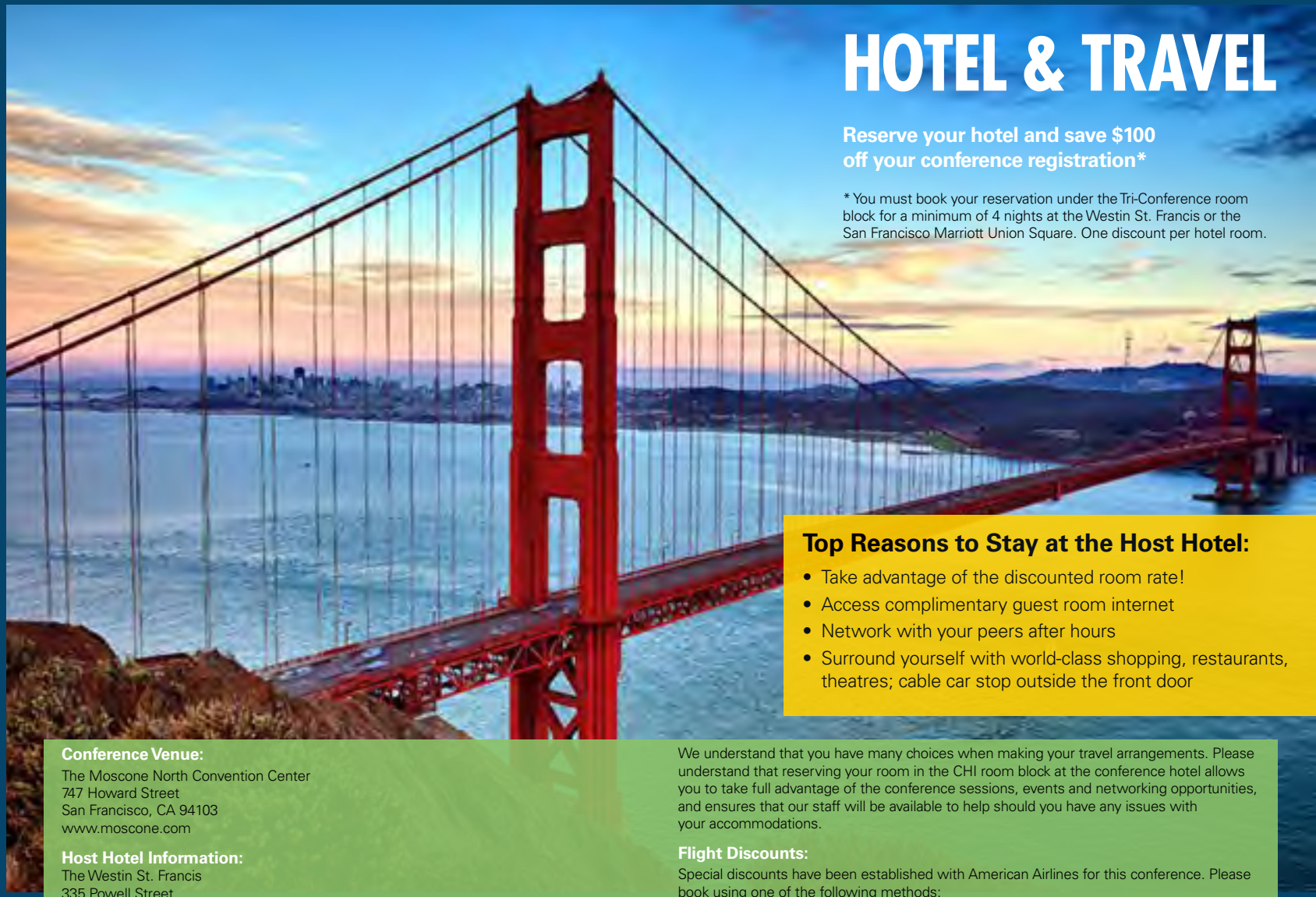

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SC1: CTCs from Bench to Bed: Streamlining from Research to Clinical Practice
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SC3: Starting an NGS Lab Part I: Technical Considerations
SC4: Sequencing 101
SC5: Best Practices in Personalized and Translational Medicine
SC6: PCR Part 1
SC7: Maximizing Reimbursement

Sunday, February 9 - 5:30 - 8:30pm

SC9: Starting an NGS Lab Part II: Practical and Business Aspects
SC10: NGS Assembly and Alignment
SC11: PCR Part II: Digital PCR Applications and Advances
SC12: Regulatory Compliance in Drug-Diagnostics Co-Development
SC13: Building an Investigational Program

Monday, February 10 - 8:30 - 11:30am

SC17: Commercialization Boot Camp
SC18: Next Generation Sequencing in Molecular Pathology
SC19: Systems Biology
SC20: Neuro-Innovation
SC21: Isolation and Characterization of Cancer Stem Cells
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