The free falling cost of whole genome sequencing raises the possibility – some say the inevitability – of applying this revolutionary technology in the clinic. Medical geneticists are now proposing the idea of a “sequence once, read often” approach to preventive medicine. However, huge challenges spanning scientific, regulatory, ethical and financial areas stand in the way of clinical genome sequencing becoming part of mainstream medicine. We invite stakeholders from all arenas impacting clinical genomics to share new findings and solutions in advancing the application of clinical genome sequencing.
American Pathologists presents significant opportunities and challenges for researchers and clinicians up a CLIA certified genomic laboratory, and what you could expect during a
an emphasis on the requirements that should be considered when setting cover the CLIA certification process, a summary of the CLIA regulations with
requirements and are subject to onsite inspections. This presentation willecome certified, clinical laboratories must be in compliance with the CLIA
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The Clinical Laboratory Improvement Amendments of 1988 (CLIA) requires
US DHHS), Centers for Medicare and
the US Department of Health and
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cover the CLIA certification process, a summary of the CLIA regulations with
an emphasis on the requirements that should be considered when setting up a CLIA certified genomic laboratory, and what you could expect during a
CLIA survey.

Development of Laboratory Standards for Next-Generation Sequencing as a Clinical Tool
Nazneen Aziz, Ph.D., Director of Molecular Medicine, Transformation Program Office, College of American Pathologists
The rapid evolution of next generation sequencing (NGS) technologies presents significant opportunities and challenges for researchers and clinicians
for improving health outcomes; particularly with respect to an increased emphasis on personalized and preventive medicine. Adoption of NGS in
the clinical laboratory setting requires the adoption of many processes and procedures, such as, the analytic and clinical validation of the test, CLIA/CAP
certification, standards for reference materials for proficiency testing, and
questions regarding reimbursement and informed consent. This talk will cover
what is practically needed for clinical adoption of NGS such as regulatory and
professional standards, development, availability, and access to reference
materials, and the laboratory professional’s role for ensuring high quality test
results that are useful for informing clinical decision making. Dr. Aziz will focus
on the laboratory accreditation standards being developed at CAP for NGS in
collaboration with ACMG and AMP.

Implementing Tools and Platforms to Support Interpretation of Clinical Diagnostic Whole Genome Sequencing Data
Elizabeth A. Worthey, Ph.D., Assistant Professor, The Human
and Molecular Genetics Center, Pediatrics, Bioinformatics, Medical College of Wisconsin
A variety of analysis tools and pipelines are required to support the clinical
interpretation of clinical diagnostic whole genome sequence data. Tools for
data tracking, pipelines for tertiary sequence analysis providing functional
data for clinical interpretation of variants, tools to identify likely deficiencies or
limitations in the data, platforms to integrate and perform QA on disparate data
types, systems to hold, organise, and query vast quantities of reference data,
and tools to support clinicians and provide visualisation to increase efficiency and
effectiveness are all required for successful clinical implementation of
WGS. This presentation will cover all of these aspects, with guidelines and
findings gleaned from 18 months of running this type of program at the
Medical College of Wisconsin: Dr. Worthey will focus on the tools and systems
required, the possible pitfalls and potential solutions in this type of analysis,
and the process of implementing these tools and pipelines within a CAP and
CLIA environment.

Clinical Interpretation of Whole Genome Sequencing
Euan Ashley, Ph.D., Assistant Professor, Cardiovascular Medicine, Stanford University School of Medicine
The cost of human genome sequencing has fallen significantly and
the availability of such information to the practicing clinician is near term. We
have recently developed a pipeline for the analysis of personal genomes
suitable for health-care applications. In this talk, I will provide examples of the
application of whole genome sequencing to the care of patients and families.

Implementing Next-Generation Sequencing at the Mayo Clinic: The Coming Revolution!
David I. Smith, Ph.D., Professor, Department of Laboratory Medicine and Pathology; Chairman, Technology Assessment Group, Center for Individualized Medicine, Mayo Clinic
I am going to review the recent developments in Next-Generation sequencing
that have resulted in dramatic increases in sequence output. As the cost for full
genome sequencing continues to drop it will herald a revolution in the practice
of medicine. These technologies can be used for so much more than whole
genome sequencing. I will then discuss how the Mayo Clinic is approaching
these technologies and the development of molecular tests based upon
Next-Generation sequencing. I will finally discuss how we are setting up the
necessary facilities to both do Next-Generation sequencing and to analyze the
resulting data.

* Separate registration is required for each Short Course

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**PRE-CONFERENCE SHORT COURSES**

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**1:30 pm Short Course Registration**

**2:00- 5:00 Short Course One**

**The Diagnostic Odyssey for Genome Sequencing**

The move to push genome sequencing into routine clinical practice requires
arguably as much skill in navigating bureaucracy as mapping and interpreting
base pairs. This short course provides practical information on crafting
clinical sequencing and analysis pipeline strategies while navigating the
route of CAP and CLIA certification by those who have sailed through these
uncharted waters.

**Instructors**

**CLIA 101: Basic CLIA Information**

Gary Yamamoto, Laboratory Consultant, United States Department of Health and Human Services (US DHHS), Centers for Medicare and Medicaid Services (CMS)
The Clinical Laboratory Improvement Amendments of 1988 (CLIA) requires
each clinical laboratories be certified by the US Department of Health and
Human Services, Centers for Medicare & Medicaid Services (CMS). To
become certified, clinical laboratories must be in compliance with the CLIA
requirements and are subject to onsite inspections. This presentation will
cover the CLIA certification process, a summary of the CLIA regulations with
an emphasis on the requirements that should be considered when setting up a CLIA certified genomic laboratory, and what you could expect during a
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on the laboratory accreditation standards being developed at CAP for NGS in
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5:00 Shared Dinner for Short Courses One and Two

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**4:30 pm Short Course Registration**

**5:30-8:30 Short Course Two**

**The Saga of Genomic Interpretation for Physicians, Payers, Patients**

To succeed, clinical genome sequencing must have the ability to enhance
medical decision making. Primary care physicians are often on the front line of
responding to "what does this mean/what do I do?" questions. Their informed
response is a saga of detailed reports including 1) physicians who order the
genomic test 2) laboratory testing personnel who run the test and generate
the data 3) bioinformaticists who prepare reports by aligning, annotating, and
analyzing the genomic data 4) molecular pathologists who interpret the data
and confirm clinical relevance 5) physicians who communicate results back to
the patients and 6) patients who respond to treatment. Step by step informed
decisions will lead to the success of the clinical genomics saga.

**Instructors**

**Delivering the Message: Translating Complex Genomic Data for Providers and Patients**

Elissa Levin, Vice President, Genomic Services, Navigenics, Inc.
One of the greatest challenges we face in the era of personalized
medicine is how to deliver complex genomic data to both providers and
patients in a manner that is accessible, understandable, and current.
Navigenics, a personalized genomic testing service that has offered testing
services through a dynamic reporting and educational platform for the past five
years, focuses on clinically relevant testing to inform clinical decision-making
and motivate behavior change. I will review our experience with physicians
and patients, as well as the inclusion of genetic counseling to facilitate the
integration of genomic data into clinical care.

**Clinical Interpretation of Whole Genome Sequencing**

Euan Ashley, Ph.D., Assistant Professor, Cardiovascular Medicine, Stanford University School of Medicine
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the availability of such information to the practicing clinician is near term. We
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interpretation capabilities of a group of highly motivated knowledgeable professionals in genetics, by sequencing their genome and providing back both the raw data and interpretation on medically relevant variants. I will present current results of this study focusing on the designed and informed consent procedure, as well as the lessons learned from various genome interpretation platforms that we provided to the participants.

6:00-6:30 Early Main Conference Registration

TUESDAY, JUNE 12

8:00 am Conference Registration & Morning Coffee

**PLENARY KEYNOTE SESSION**

Translational Genomics: Bridging the Gap between Bench and Bedside

8:35 Chairperson’s Opening Remarks

8:45 Lessons from Whole Genome Sequencing in the Clinic
Howard J. Jacob, Ph.D., Professor and Director, Human and Molecular Genetics Center, Medical College of Wisconsin

In November of 2010, the Medical College of Wisconsin (MCW) and Children's Hospital and Health System (CHHS) implemented a clinic offering whole genome sequencing (WGS) to end diagnostic odysseys. Our data generation (sequencing) and data analysis operate under a CAP/CLIA clinical laboratory environment. We have had 50 clinical cases nominated and 14 have undergone clinical sequencing and analysis. In our estimation, the biggest limitation for our program is the availability of WGS to compare to. As more genomes become available, and more clinics bring sequencing online, there will be expanded clinical utility. Pharmacogenomics and cancer are likely other clinical conditions that would benefit from WGS today.

9:30 Whole Genome Analysis: Efficacy, Accuracy, and Application in Best Practice Care
Peter J. Tonellato, Ph.D., Director, Laboratory for Personalized Medicine, Center for Biomedical Informatics, and Professor (visiting), Department of Pathology, Beth Israel Deaconess Medical Center and Harvard Medical School

The Department of Pathology, Beth Israel Deaconess Medical Center has joined with the Center for Biomedical Informatics, Harvard Medical School to work with a consortium of stakeholders from health care, government and private industry to test the use of whole genome sequencing in genome-era medicine. Much like that created in biomedical science, whole genome sequencing, expression profiles and other high-throughput technologies will create a post-genome paradigm shift in health, disease prevention, and personalized medicine. However, this paradigm shift will not take place until the technologies and processes are re-engineered for use in the research setting into use in the best-practice care delivery setting. Our efforts include bioinformatics tools, methods and analysis to process and integrate personal genomes into the EHR, redefining regulatory management of whole genome sequencing in the clinical laboratory, reimbursement, and creating a robust clinical business model for post-genome pathology practice. These and parallel efforts will catalyze the adoption and widespread implementation of the post-genome competency required to fully capture the value of whole genome information and thereby position the discipline of pathology to lead rather than follow in the coming era of personalized medicine.

10:15 Coffee Break, with Exhibit and Poster Viewing

**Trends in Genome Interpretation**

10:45 Data-Driven Personalized Medicine
Atul Butte, M.D., Ph.D., Chief, Division of Systems Medicine, Department of Pediatrics; Associate Professor, Stanford University School of Medicine; Director, Center for Pediatric Bioinformatics, Lucile Packard Children’s Hospital

Dr. Butte, a bioinformatician and pediatric endocrinologist, will highlight his lab’s work on environment-wide association studies, evolution and disease, clinical evaluations of patients presenting with whole genome sequences, and gene-environment interactions to better understand the nature of disease susceptibility and treatment.

11:30 Pathology and the Third Wave of Medical Genomics
Mark Boguski, M.D., Ph.D., Associate Director, Pathology, Center for Biomedical Informatics, Harvard Medical School

12:15 pm Close of Morning Session

12:30 Luncheon Presentation
Whole Genomes Take Root in Clinical Intelligence

Nathan Pearson, Ph.D., Director, Research, Knome

As sequencing costs drop, reference data grow, and interpretation methods sharpen, the clinical utility of whole genomes is spreading beyond its first footholds in pediatrics and oncology. Taking full advantage of these advances, the knomeCLINIC early technology access program offers clinics a clear path to installing smart, robust genome interpretation pipelines that generate clinically decisive insights and integrate them seamlessly into patient care.

**PLENARY KEYNOTE SESSION (CONT.)**

Genomic Medicine: Translating into the Clinic

1:45 Chairperson’s Remarks

1:50 Translational Genomics and Health Outcomes: The Path to Genomic Medicine
Robert C. Green, M.D., MPH, Associate Director for Research, Partners Center for Personalized Genetic Medicine, Division of Genetics, Department of Medicine, Brigham and Women’s Hospital and Harvard Medical School

The successful integration of genomic science in clinical medicine will involve the generation and disclosure of risk information on a scale not previously encountered in medicine. There is an urgent need for empirical research studies that combine principles of epidemiology, clinical trials and comparative effectiveness to illuminate best practices, and examine the behavioral and health outcomes associated with using probabilistic genetic information. We have called this new research discipline Translational Genomics and Health Outcomes. In this presentation, a number of completed and ongoing research studies in this area will be highlighted, emphasizing the lessons learned and the challenges to come in this new field. The presentation will also preview the recently funded MedSeq™ Project, the first randomized controlled trial to examine the impact of whole genome sequencing in the practice of medicine.


Investigating the genetic basis of disease or drug response

Building new software and cloud-based analysis pipelines

Interpreting genetic variations

Transferring raw sequencing data

Integrating with other clinical data systems

Creating new medical-grade databases

Reporting relevant clinical information in a physician-friendly manner

Continuous learning feedback

Cypher Genomics has developed a unified framework to facilitate genome interpretation in both discovery and pathology settings using a combination of predictive and prior-knowledge-based variant annotations, as well as a suite of tools used to filter and prioritize genomic variants. We will describe real examples of the application of these tools to disease variant discovery in idiopathic disease cases.

Sultan Meghji, Vice President, Product Strategy, Appistry

With Ayrris™ / BIO, Appistry brings intelligence to the process of moving, analyzing and storing genetic data— and a proactive approach to genomic intelligence and personalized medicine. In this software spotlight, Sultan Meghji will cover HPC and Cloud technologies that empower users to integrate clinically actionable bioinformatics into their practice by managing the complexities of NGS analysis and data management, while remaining compliant with regulatory structures. Case studies included.

Sultan Meghji, Vice President, Product Strategy, Appistry

Even more exciting is the way that crowds of genetically characterized individuals are coming together to contribute anonymized information about their own health histories. By combining phenotypic with genotypic data, researchers are accelerating the search for breakthroughs that could have exceptional personal relevance to the individual participants who enable those discoveries. 23andMe has defined this new paradigm of crowd-sourced genetic discovery, transforming Research ➝ MeSearch ➝ WeSearch. Celebrities including Martha Stewart, Ivanka Trump, Mehmet Oz, and Muhammed Ali have joined to discover things about themselves or to advance research. And companies like Genentech, Johnson & Johnson, and Google have backed the company to help it achieve its ambitious goals.

Ashley Dombkowski, Ph.D., Chief Business Officer, 23andMe, Inc.

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Today, for a few hundred dollars, a consumer can get an in-depth look at a million bits of his/her personal genetic code along with explanations of what these things might mean. From fun things like deep genetic ancestry or a personal predisposition to sneeze in bright sunlight to serious things like: do genetics increase her risk for blood clots if she takes oral contraceptives; or is she a carrier of a disease like cystic fibrosis.

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3:10 Talk Title to be Announced

Ashley Dombkowski, Ph.D., Chief Business Officer, 23andMe, Inc.

Genetics, Consumers, and the Wisdom of Crowds

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3:35 Refreshment Break, with Exhibit and Poster Viewing

4:15 Human Genome Interpretation

Ali Torkamani, CSO, Cypher Genomics

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4:35 Intelligent Genomics in the Cloud

Dietrich A. Stephan, Ph.D., CEO, Silicon Valley Biosystems (SVBio)

Every human disease has a genetic component and NGS promises broad-based delivery of actionable clinical information to benefit outcomes and reduce costs of care. While promising, NGS technologies and data are complex, creating new pain-points for the consumers of the ultimate data. SVBio’s solution is scalable turn-key diagnostic service to enable NGS-based MDx to flourish and impact patients, physicians and payors positively.

Additional Presentation Opportunities Available

5:15 NGS Variant Analysis for Clinical Applications: High Quality, Reproducible, Standardizable Workflows Required!

Peter Grant, CEO, Genomatix Software, Inc.

NGS technologies produce very rich data sets. While the wet-lab portion of many applications has reached a level of maturity, data analysis remains the bottleneck for a real return on investment. In addition, clinical application of NGS driven analyses require a high degree of clarity of results and reproducible workflows that can be standardized. Genomatix provides a high-end technology in an easy to use platform for such applications. An example will be presented.

5:35 Welcome Reception with Exhibit and Poster Viewing

6:35 Close of Day
Concurrent Breakout Sessions

Roads to Regulation & Certification

8:30 Chairperson’s Remarks

8:35 Implementation of Next-Generation Sequencing as a Clinical Test

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

The technological advancements in next-generation sequencing have been astounding. The cost of sequencing is now at a price-point where it is conceivable that the tests for genome, exome and targeted gene panel analysis will be offered in many diagnostic laboratories. The College of American Pathologists has convened a group of its member experts including representatives from the Association of Molecular Pathology and the American College of Medical Genetics to develop standards for next generation sequencing as a clinical test. Dr. Aziz will address some of the standards that are being considered by the workgroup for the wet bench workflow and bioinformatics pipeline and the approach that CAP is taking for implementing these standards for this rapidly evolving technology.

9:05 Impact of Direct-to-Consumer Tests

9:35 Ethical, Legal, and Practical Challenges in Follow-Up

John West, CEO, Personalis, Inc.

The cost of DNA sequencing has dropped dramatically in recent years but accuracy and interpretation remain challenges. Sequencing families can help. I report our experience as the first family of four sequenced by Illumina (in late 2009). I will also outline advances in genome interpretation technology at our new company, Personalis, Inc. In our experience, genome sequencing is useful today, but much work remains to be done.

9:05 Impact of DTC Genomic Testing at Long-Term Follow-Up

Cinnamon S. Bloss, Ph.D., Assistant Professor, Scripps Translational Science Institute, Scripps Genomic Medicine

Direct-to-Consumer (DTC) genome-wide disease risk profiling is currently available to individuals and provides information about one’s genetic risk for a range of common polygenic diseases. The Scripps Genomic Health Initiative (SGHI) was launched in 2008 with the aim of evaluating the psychological, behavioral, and clinical impacts of DTC genomic testing on consumers. In a recently published study of over 2,000 individuals who underwent genomic profiling with the Navigenics Health Compass, we found minimal psychological, behavioral, or clinical impacts of testing at short-term follow-up. We will present new data based on long-term follow-up of the same group of consumers. Overall, findings from these new data are similar to those from short-term follow-up and suggest a general lack of impact of testing with respect to the outcomes assessed. Analyses evaluating the impact of disease risk testing for relevant subgroups of individuals, including those with or without a family history of disease, as well as younger versus older individuals will also be presented. Preliminary data on the impact of DTC pharmacogenomic testing, as well as genetic ancestry testing will also be presented. Finally, we will discuss new directions for research in consumer genomic testing.

9:35 Operational Challenges of Implementing Clinical Genomics in Genetics Laboratories

Mike M. Moradian, Ph.D., Director of Operations / Molecular Genetics Scientist, Kaiser Permanente Southern California Regional Genetics Laboratory

Clinical Genomics is a recent concept in today’s complex and dynamic Molecular Pathology laboratory set up, which requires detailed operational evaluation. Laboratories could face many challenges when deciding to implement Clinical Genomics. These challenges may include: instrumentation, testing platforms, technologist training, assay validation, result generation, bioinformatics expertise in result interpretation, and preparation of an appropriate report for physician(clinic use). Of course regulatory, compliance, and billing/reimbursement issues obligate comparable and necessary action. A brief description of these operational challenges and possible solutions will be discussed.

Impact of Direct-to-Consumer Tests

8:30 Chairperson’s Remarks

8:35 Sequencing in the Family

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Issues with Returning WGS Results

9:35 Ethical, Legal, and Practical Challenges in Clinical Practice

Hank Greely, J.D., Deane F. and Kate Edelman Johnson Professor of Law; Director, Center for Law and the Biosciences; Professor (by courtesy) of Genetics, Stanford School of Medicine; Chair, Steering Committee of the Center for Biomedical Ethics; and Director, Stanford Interdisciplinary Group on Neuroscience and Society

The sequencing and analysis of patients’ whole genome will present substantial technical and scientific challenges. More difficult, however, will be the ethical, legal, and practical challenges, in sequencing, in interpretation, and, most troubling, in returning the results in a useful way to patients. This talk will discuss those issues.

10:05 Clinical Issues with the Return of Next-Generation Sequencing Results

Gholson J. Lyon, M.D., Ph.D., Adjunct Assistant Professor, Child and Adolescent Psychiatry, New York University

Prepare Now for Reimbursement

9:35 Operational Challenges of Implementing Clinical Genomics in Genetics Laboratories

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10:05 Reimbursement: How Health Plans Evaluate New Technology
Gary M. Owens, M.D., Gary Owens Associates
Health plans are faced with reimbursement issues regarding new and emerging technology on a regular basis. Because plans must manage their client's resources wisely, they must balance allowing appropriate access to effective new technology with the potential to waste resources on technologies that are either an inefficient use of resources or clinically ineffective. Plans have developed rigorous evidence-based processes to evaluate new technology. Failure to understand the evidentiary requirements of a health plan can lead to delays in reimbursement or failure to obtain reimbursement at all for new technologies. This session will provide insight into the technology evaluation process and how plans look at clinical evidence, comparative effectiveness and cost when they review new technologies.

10:35 Coffee Break, with Exhibit and Poster Viewing

11:15 PLENARY PANEL DISCUSSION
Genomic Medicine: The Road Ahead
There is sharply growing momentum for the introduction of genome sequencing and analysis in clinical medicine, especially with the much hyped ‘$1,000 genome’ poised to arrive in 2013. But while there have been some gratifying case reports of the diagnostic and therapeutic potential of clinical genome sequencing, they only serve to emphasize the daunting challenges that must be tackled for genomic medicine to reach its potential, including issues surrounding interpretation and education, counseling and consent, regulation and reimbursement. In this open, interactive roundtable, a group of speakers frankly debate the road ahead for genomic medicine.

Moderator: Kevin Davies, Ph.D., Chief Editor, Bio-IT World
Panelists: Hank Greely, J.D., Deane F. and Kate Edelman Johnson Professor of Law, Director, Center for Law and the Biosciences; Professor (by courtesy) of Genetics, Stanford School of Medicine; Chair, Steering Committee of the Center for Biomedical Ethics; and Director, Stanford Interdisciplinary Group on Neuroscience and Society
Grant Campany, Senior Director, Prize Lead, X Prize Foundation
Linda Avey, Co-Founder and CEO, Curious, Inc.
Stefan Roever, CEO & Founder, Genia Technologies
Andy Watson, Chief Marketing Officer, RainDance Technologies, Inc.
Richard Resnick, CEO, GenomeQuest, Inc.

12:15 pm Close of Morning Session

12:30 Luncheon Presentation
Solving the Challenges of DNA Sequencing for Personal Diagnostics
Stefan Roever, CEO & Founder, Genia Technologies
There is no debate that genetic information is needed to truly realize the promise of personalized medicine. The problem is that today’s DNA sequencers cost anywhere from $50K - $1M, rely on complicated optics, and utilize a complex workflow that does not lend itself to clinical utility. Genia is using standard semiconductor technology to enable massively parallel, single molecule DNA sequencing. The company has developed a versatile nanopore-based platform which allows for single molecule, electrical, real-time analysis.

Child Study Center; Research Scientist Center for Applied Genomics, Children’s Hospital of Philadelphia and Utah Foundation for Biomedical Research
I would like to present here two real-world sequencing scenarios that I have encountered during my research, which I believe highlight some major issues facing the research community currently engaged in the sequencing of human genomes. I believe that it is imperative that exome and whole genome sequencing of humans be performed in environments that allow ready dissemination of these data back to the people donating their samples. This means performing the collection of samples and the subsequent sequencing as rigorously as with any other clinical lab test, so that all data can be returned easily to research participants. I will discuss the clinical, ethical and practical aspects of trying to achieve this.

10:35 Coffee Break, with Exhibit and Poster Viewing

PLENARY SESSION
Trends in Genetic Diagnostic Platforms

1:30 Chairperson’s Remarks

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

1:35 Whole Genome Sequencing as a Clinical Tool
Jill Hagenkord, M.D., Chief Medical Officer, Complete Genomics
Whole genome sequencing has the potential to impact healthcare in the near future. This talk will highlight examples of cost-effective utilization of this powerful tool in cancer and constitutional diagnostics, as well as foreshadow the impact of WGS in wellness and disease applications.

2:05 Technology Showcase
The Clinical Exome: Diagnosing Genetic Disease Today
Elizabeth Chao, Ph.D., M.D., Director of Translational Medicine at Ambry Genetics
The Clinical Diagnostic Exome™ has expanded the landscape of medical genetics and rapidly translated into a clinically useful tool. Nearly all known disease-causing mutations in humans today are found within exons or protein-coding regions the exome captures, the best understood and most functionally relevant (to date) regions of the genome. This session will focus on the success as well as future challenges of clinical exome testing, from the first company to launch exome testing into the clinical space.

2:35 Speed Cures: Genomic Information Enters Clinical Medicine
Paul R. Billings M.D., Ph.D., CMO, Life Technologies
Except in dire emergencies, medicine has been historically slow and deliberative. The physician, striving to be a compassionate healer, was not hurried. The resulting system of care was for many ineffective, costly, slow, error prone and an “odyssey”. The rapid availability of basic, high quality genomic information is transforming healthcare provision and will alter many practices. This talk will highlight clinical care issues that are evolving as democratized comprehensive genetic analysis enters medical care.

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3:05 Defining the Complete Solution for Clinical Interpretation of a Whole Genome Sequence

Tina Hambuch, Ph.D., Senior Scientist, Illumina Clinical Services Laboratory, Illumina, Inc.

Platform advances have enabled greater access to whole genome sequencing by the clinical community however extracting the full medical value of this information remains a community challenge. Certified since 2009, the Illumina Clinical Laboratory is the only CLIA/CAP service for whole genome sequencing. In this talk, Illumina will discuss a workflow for clinical interpretation and reporting, the need for a clinical grade database, and introduce the MyGenome app for visualization and communication of clinical interpretations.

3:35 Refreshment Break, Last Chance for Exhibit and Poster Viewing

4:00 Progress in Building a Comprehensive Information and Technology Infrastructure for Whole Transcriptome RNA-Seq and Targeted Mutation Analysis across Large Populations of Cancer Patients

Steven Shak, M.D., Chief Medical Officer, Genomic Health

As a society, we spend billions of dollars treating diseases we don’t understand – resulting in large misallocations of resources and poor outcomes. Massively parallel sequencing is a powerful new tool for redefining disease at the molecular level and transforming the way medicine is practiced. This talk will center around the ability to scale whole transcriptome RNA-seq and targeted mutation analysis on fixed paraffin-embedded tissues across large populations of cancer patients with the goal of merging the healthcare system and the clinical trial system to accelerate our understanding of disease and facilitate cures.

4:30 Where the Rubber Meets the Road – Translating Next-Generation Sequencing from a Research Application to a Standardized Lab-Developed Test in a CLIA Lab

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

Next-generation sequencing (NGS) has transformed the research sequencing landscape. These technologies have enabled a plethora of new discoveries by allowing us to answer questions more cost-effectively than in past and ask questions that were impossible to address by capillary electrophoresis sequencing or high density chip platforms. Translating NGS applications from a research environment to a standardized test offering in a clinical reference laboratory presents multiple challenges. These range from identifying NGS tests that provide clinical value beyond existing tests on traditional platforms, increased need for a bioinformatics infrastructure to interpret and report results, genome sequence versioning and subscription access to permit revisiting a prior sequence because of the identification of new indications, to consequences of updates to the technology hardware and software as well as reagents of the rapidly changing NGS field. Changes in the regulatory landscape that may impact the use of these technologies are also being contemplated. This presentation will offer a users perspective of these issues.

5:00 Panel Discussion with Afternoon Speakers

5:30 Close of Conference
SPONSOR & EXHIBITOR OPPORTUNITIES

CHI offers comprehensive sponsorship packages which include presentation opportunities, exhibit space and branding, as well as the use of the pre and post show delegate list. Sponsorship allows you to achieve your objectives before, during, and long after the event. Any sponsorship can be customized to meet your company’s needs and budget. Signing on earlier will allow you to maximize exposure to hard-to-reach decision makers.

Sponsored Presentations
Showcase your solutions to a guaranteed, highly-targeted audience. Package includes a 15 or 30-minute podium presentation within the scientific agenda, exhibit space, on-site branding and access to cooperative marketing efforts by CHI.

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Opportunity includes a 30-minute podium presentation. Boxed lunches are delivered into the main session room, which guarantees audience attendance and participation. A limited number of presentations are available for sponsorship and they will sell out quickly. Sign on early to secure your talk!

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Sponsors will hand-pick their top prospects from the conference pre-registration list for an evening of networking at the hotel or at a choice local venue. CHI will extend invitations and deliver prospects. Evening will be customized according to sponsor’s objectives (i.e. purely social, focus group, reception style or plated dinner, plated dinner with specific conversation focus).

User Group Meeting
Take advantage of the prestigious audience in attendance to conduct market research or gather feedback on your new product or service on-site. CHI will provide a meeting room set for 50-75 delegates, equipped with AV including an LCD screen. This presents a rare opportunity to meet with a large, targeted group of end-users, and walk away from the conference with qualified leads and information!

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Includes a 15-minute or 30-minute podium presentation during the pre-conference workshop, as well as your company logo displayed on pre-conference workshop materials and on-site signage.

CHI Lead Generation
CHI can help you with lead generation throughout the year. Our internal database includes over 800,000 prospects in the life sciences. By leveraging the database and mining for your specific requirements, we can produce multiple custom projects which will deliver your prospective buyers: Web Symposiaums, Podcasts, White Papers, Custom Market Research Surveys and more!

Exhibit Information
Exhibitors will enjoy facilitated networking opportunities with qualified decision makers at TCGC: The Clinical Genome Conference, making it the perfect platform to launch a new product, collect feedback and generate new leads. Exhibit space sells out quickly, so reserve yours today!

Additional networking and promotional opportunities are available!

To discuss the various ways your company can participate as a sponsor or exhibitor, please contact:

Jon Stroup
Business Development Manager
781-972-5483 | jstroup@healthtech.com

HOTEL & TRAVEL INFORMATION

Conference Hotel:
Hotel Kabuki
1625 Post Street
San Francisco, CA 94115
Phone: 415-922-3200
Reservation Line: 800-533-4567

Discounted Room Rate: $140 s/d
Discounted Room Rate Cut-off Date: May 14, 2012

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

Flight Discounts:
Special discounts have been established with American Airlines for this conference.
- Call American Airlines 1-800-433-1790 and use Conference code 7162AX.
- Go to www.aa.com/group and enter Conference code 7162AX in promotion discount box.
- Contact our dedicated travel agents at 1-877-559-5549 or chi@protravelinc.com.

Car Rental Discounts:
Special rental discounts have been established with Hertz for this conference.
- Call Hertz 1-800-654-3131 and use our Hertz Convention Number (CV): 04KL0003
- Go to www.hertz.com and use our Hertz Convention Number (CV): 04KL0003
Pricing and Registration Information

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<tr>
<td>Advance Registration Discount until May 11, 2012</td>
<td>$1495</td>
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<td>Registrations after May 11, 2012, and on-site</td>
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**PRE-CONFERENCE SHORT COURSE PRICING (JUNE 11)**

- Single Short Course: $645 ($325)
- Two Short Courses: $945 ($625)

**CONFERENCE DISCOUNTS**

- Poster Submission-Discount ($50 Off): Poster abstracts are due by May 17, 2012. Once your registration has been fully processed, we will send an email containing a unique link allowing you to submit your poster abstract. If you do not receive your link within 5 business days, please contact jring@healthtech.com. * CHI reserves the right to publish your poster title and abstract in various marketing materials and products.

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

Additional discounts are available for multiple attendees from the same organization. For more information on group rates contact David Cunningham at +1-781-972-5472.

How to Register: healthtech.com/CLG

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Please refer to the Registration Code below:

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