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Keynote Presentations

Consumers in the Lab and the Clinic: My Daughter's DNA...and Every Little Thing About Her

Hugh Young Rienhoff, Jr., M.D., Director, MyDaughtersDNA.org

Consumer Genomics: From Niche to Mainstream

Spencer Wells, Ph.D., Explorer-in-Residence and Director, The Genographic Project, National Geographic Society

5th Annual

CONSUMER GENETICS CONFERENCE

September 25-27, 2013

Seaport World Trade Center | Boston, MA

Empowering Patients and Consumers with Advances in Genomics, Diagnostics and Personalized Healthcare

Program Topics:

- Whole Genome Debates
- Translational Genomics
- Clinical & Third-Generation Sequencing
- Personal Genome Analysis & Interpretation
- Empowering Patients: Companies & Technologies
- Molecular Diagnostics & Point-of-Care
- Investment & Funding Opportunities
- Reimbursement Models
- Five-Year Plan for Consumer Genomics
- Data Analysis & Management
- Ethics, Privacy & Regulation
- Digital Health Tracking Apps

The Consumer Genetics Conference (CGC) is a one-of-a-kind event that draws together a dynamic community of scientists, clinicians, technology innovators, and patients to discuss the burning issues around the analysis and delivery of genomics results as presented directly to patients and consumers.

Over three days, attendees will hear about disruptive diagnostic technologies, cognitive barriers to patients (and medical professionals), ethical/regulatory/privacy issues, the thorny issue of reimbursement, and the challenges of building relationships to realize the potential of personal genomics and individualized medicine. CGC provides an opportunity for all stakeholders to come together at one venue, share viewpoints and engage in an honest dialogue, and together learn how to move the elephant of change.

Corporate Sponsors

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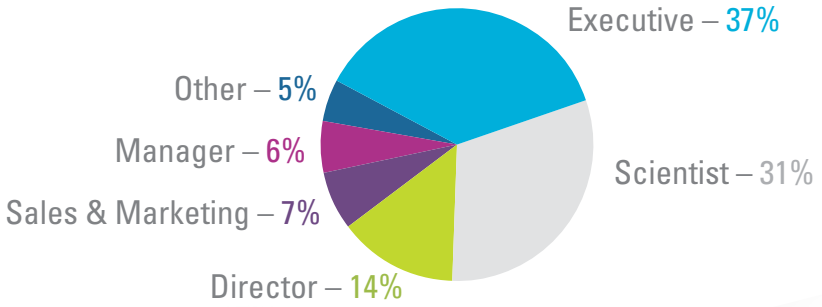
ConsumerGeneticsConference.com

Event History

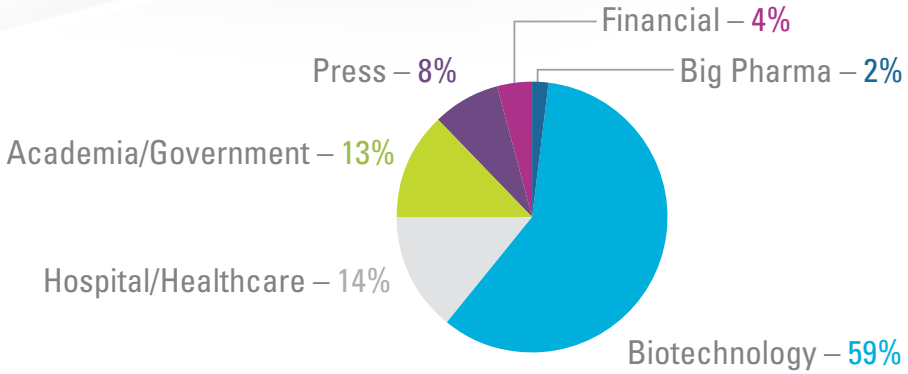
Since its debut in 2009, the annual Consumer Genetics Conference (CGC) has established itself as the original, unbiased event for advancing personal genomics to empower individualized healthcare. The program features business, academic, medical, and technology thought leaders presenting the latest research and case studies on the progress of an individual's ability to investigate their own DNA sequence, whether for health, genealogy or just plain curiosity, and the broader ecosystem behind advances in DNA sequencing, diagnostics, and forms of personalized healthcare. CGC provides a unique outlet where all voices can be heard: pro & con, physician & consumer, research & clinical, academic & corporate, financial & regulatory.

Attendee Titles



Who Attends

The Consumer Genetics Conference brings together over 500 thought leaders and stakeholders involved in the ecosystem of delivering high-value genetics and genomics results directly to patients and consumers. Stakeholders include: academic research labs and medical centers, sequencing technology companies, information technology service providers, clinical/hospital laboratories, regulators, investors, lawyers, genetic counselors, health care practitioners, patient advocacy groups, and the press.



Organization Type

For questions or suggestions about the meeting, please contact:

Cindy Crowninshield, RD, LDN, Conference Director
781-354-0120 | cindy@consumer-genetics-conference.com

For sponsorship and exhibit sales, information including podium presentations, contact:

Jay Mulhern, Manager, Business Development, Conferences & Media
781-972-1359 | jay@consumer-genetics-conference.com

"Concise and well run meeting on a field that is (finally) about to take off."
CEO, CBT Advisors

"Excellent conference – a must for next-generation sequencing pioneers."
Fellow, Pediatric Genetics, Children's Hospital Boston

Sponsorship, Exhibit, and Lead Generation Opportunities

Comprehensive sponsorship packages are available for your company to promote its products and/or solutions directly to organizations empowering patients and consumers with advances in genomics, diagnostics and personalized healthcare. Customizable sponsorship packages allow you to achieve your objectives before, during, and long after the event. Signing on earlier will allow you to maximize exposure to hard-to-reach decision makers!

Agenda 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.

Breakfast & Luncheon Presentations

Opportunities include 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 to secure your talk!

Invitation-Only VIP Dinner/Hospitality Suite

Sponsors will select their top prospects from the conference pre-registration list for an evening of networking at the hotel or at a choice local venue. We 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).

Exhibit Information

Exhibitors will enjoy facilitated networking opportunities with qualified decision-makers at the Consumer Genetics 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 promotional opportunities are available including:

- Conference tote bags
- Badge lanyards
- Tote bag inserts of company literature
- Padfolios
- Keynote chair drop of company literature
- Session room chair drop of company literature
- Program guide sponsor
- Poster abstract book sponsor

Looking for additional ways to drive leads to your sales team? We can help through:

Custom Lead Generation Programs:

- Targeted campaign promotion to unparalleled database of 800,000+ individuals in the life sciences
- Experienced marketing team promotes campaign, increasing awareness and leads

Live Webinars:

- Assistance in procuring speakers
- Experienced moderators
- Dedicated operations team to coordinate all efforts

Whitepapers:

- Industry recognized authors, with vast editorial experience, available to help write your whitepaper

We also offer market surveys, podcasts, and more!

For sponsorship & exhibit information, please contact:

Jay Mulhern
Business Development Manager
781-972-1359
jay@consumer-genetics-conference.com

"Very informative meeting with good exposure of new technologies in the field."
CSO, ChemGenes Corp.

"On the frontier, forging the future together."
Professor, Virginia Tech

Conference-at-a-Glance

Tuesday Sept 24 1:30-4:30 pm	Pre-Conference Short Courses* Data Analysis/Management Clinical Translation of Genetic Advances Genomics 101
Tuesday Sept 24 5:00–8:00 pm (Dinner)	Dinner Short Courses*: NGS for Clinical Testing Management of Billing & Reimbursement for Molecular-Based Tests
Wednesday Sept 25	Opening Plenary Keynote Science/Research Updates on Genomics/Sequencing from Academic Medical Centers Science/Research Updates on Genomics/Sequencing from the Clinic The Funding Debate: What Insurers/Payors are Willing to Pay vs What Investors are Willing to Fund Welcome Reception with Exhibit and Poster Viewing Networking Party
Thursday Sept 26	Technology Company/Provider Spotlight: Current Trends and Visions Communicating Genetics and Science to Everyone Consumer Testing – Points and Counterpoints Impact of Patent Law on the Future of Genetic Testing Networking Party
Friday Sept 27	Digital Consumer Healthcare: Health Tracking Applications, Demos & Case Studies Consumer Landscape: Current Opportunities and the Big Picture Closing Plenary Keynote

Pre-Conference Short Courses* Tuesday, September 24, 2013

Join your colleagues for more in-depth, focused learning at an informational and interactive Short Course. Choose one of the Short Courses below for three hours of instruction and discussion in a small group setting. Get your questions answered, network with colleagues, and share ideas. See website for further details.

Afternoon Courses | 1:30 – 4:30 pm

Short Course 1: Data Analysis/Management

Caleb J Kennedy, Ph.D., Lead Scientist, Good Start Genetics, Inc.
Vasisht Tadigotla, Ph.D., Bioinformatics Scientist, Courtagen Life Sciences, Inc.

Short Course 2: Clinical Translation of Genetic Advances

Elicia Estrella, MS, CGC, LGC, Genetic Counselor and Study Coordinator, Program in Genomics/Harvard Neuromuscular Disease Project, Children's Hospital, Boston
Amanda Gammon, MS, CGC, Licensed Genetic Counselor, Huntsman Cancer Institute, University of Utah
Wendy Kohlmann, MS, CGC, Licensed Genetic Counselor, Huntsman Cancer Institute, University of Utah
Ellen T. Matloff, MS, Research Scientist, Department of Genetics and Director, Cancer Genetic Counseling, Yale Cancer Center
Erica Ramos, MS, CGC, Clinical Genomics Specialist, Illumina, Inc.
Tricia See, ScM, CGC, Genetic Counselor, Informed Medical Decisions

Short Course 3: Genomics 101

Shawn C. Baker, Ph.D., CSO, BlueSEQ
Additional Instructors to be Announced

Dinner Courses | 5:00 – 8:00 pm

Short Course 4: NGS for Clinical Testing

Nazneen Aziz, Ph.D., Director, Molecular Medicine, Transformation Program Office, College of American Pathologists
Birgit Funke, Ph.D., FACMG, Assistant Molecular Pathologist and Director, Clinical Research and Development, Laboratory for Molecular Medicine, Massachusetts General Hospital; Assistant Professor in Pathology, Harvard Medical School

Short Course 6: Management of Billing & Reimbursement for Molecular-Based Tests

Bonnie Ancone, Vice President, Molecular Diagnostics, XIFIN, Inc.
Rina Wolf, Vice President, Commercialization Strategies, XIFIN, Inc.
Kyle Fetter, Associate Vice President, Molecular Diagnostics, XIFIN, Inc.

**Separate registration is required*

Day 1: Wednesday, September 25

8:30 am Opening Remarks
John Boyce, President and CEO, GnuBIO
Meredith Salisbury, Senior Consultant, Bioscribe

8:45 Consumers in the Lab and the Clinic: My Daughter's DNA...and Every Little Thing About Her
Hugh Young Rienhoff, Jr., M.D., Director, MyDaughtersDNA.org
When his daughter was born with an undescribed syndrome, clinical geneticist and biotech entrepreneur Hugh Rienhoff launched a years-long quest to find the answer himself. In 2013, he published the results of those efforts, reporting a mutation in TGFB3 that appears to be the root of his daughter's syndrome. In this keynote address, Dr. Rienhoff will offer his views on personalized medicine and genetic-based diagnostics from the perspective of a patient's father, a clinician, and a member of the biotech industry.

9:30 Coffee Break with Exhibit & Poster Viewing

Science/Research Updates on Genomics/Sequencing from the Clinic

10:15 Chairperson's Remarks

10:20 Customer Reactions to Consumer Genetics Services
Robert Green, M.D., MPH, Associate Professor of Medicine, Division of Genetics, Department of Medicine, Brigham and Women's Hospital and Harvard Medical School
The Impact of Personal Genomics (PGen) Study is a collaboration between researchers at Brigham and Women's Hospital, the University of Michigan School of Public Health, and two genetic testing companies, 23andMe and Pathway Genomics. 1,740 new customers of these companies were recruited for the survey-based study at the time they submitted DNA samples. Goals of the study are to measure the extent to which the users understand the genetically-based risk information and to assess how consumer genetic testing influences users' risk perceptions, psychological wellbeing, health behaviors, healthcare utilization, and communication about results. This is the first prospective study of consumer genetics in which academic researchers are obtaining participants' genetic risk results. Researchers will thus be able to compare actual genetic information seen by consumers with risk perceptions and psychological and behavioral outcomes.

10:40 Supporting Genomic Medicine through Data Sharing
Heidi L. Rehm, Ph.D., FACMG, Chief Laboratory Director, Molecular Medicine, Partners HealthCare Center for Personalized Genetic Medicine (PCPGM); Assistant Professor of Pathology, Harvard Medical School
With clinical sequencing rapidly expanding, data interpretation has become the bottleneck. This talk will present approaches to share data in support of community-based approaches to advancing our knowledge of genomics and its application to medicine.

11:00 From Massive-Scale Genomic Data to Functional Interpretation
Daniel MacArthur, Ph.D., Assistant Professor, Massachusetts General Hospital; Co-founder, Genomes Unzipped
Accurately interpreting the variation present in any individual's genome requires placing it in the context of tens of thousands of other genomes - but these genomes must be accurately and consistently analyzed. This presentation describes the combined analysis of exome sequencing data from over 50,000 individuals, and the ways in which the resulting information can be used to more accurately identify disease-causing mutations in patient genomes. Recent work will be discussed on the use of deep RNA sequencing and whole-genome sequencing data in the diagnosis of rare, severe diseases.

11:20 Q&A/Discussion

11:30 Luncheon Presentation *(Sponsorship Opportunity Available)* **or Lunch on Your Own**

Science/Research Updates on Genomics/Sequencing from Academic Medical Centers

1:00 pm Chairperson's Remarks
Catherine Brownstein, Ph.D., Project Manager, The Gene Partnership, Boston Children's Hospital; Instructor, Pediatrics, Harvard Medical School

1:05 Lessons from CLARITY — An International Competition on Interpreting and Reporting Clinical Genomic Information
Catherine Brownstein, Ph.D., Project Manager, The Gene Partnership, Boston Children's Hospital; Instructor, Pediatrics, Harvard Medical School
CLARITY stands for Children's Leadership Award for the Reliable Interpretation and appropriate Transmission of Your genomic information. This international competition was launched in January 2012 with the goal of surveying current practices in bioinformatic analysis, interpretation and reporting of next-generation sequencing to diagnose rare genetic conditions. The Challenge provided contestants with DNA sequences and clinical data from three families with rare conditions for which no genetic cause had been identified. This presentation will present these results and summarize and discuss some of the conclusions and lessons from this competition.

1:25 Optimizing Analysis Pipelines for Improved Variant Discovery from Personal Genomes
Gareth Highnam, Research Scientist, David Mittelman Laboratory, Genetics and Genomic Medicine Laboratory, Virginia Polytechnic Institute
Although largely based on genotyping at the present time, DTC companies will be migrating to full sequencing very soon – whether panels or exomes or whole genomes. A major pain point is developing and evaluating analysis pipelines for analyzing next-generation sequencing data to maximize accurate detection of variants. The audience will be exposed to challenges, solutions, and opportunities that relate to the transition to next-generation sequencing in genetic testing and diagnostics.

1:45 Genetic Diagnostics Using Next-Generation Sequencing: The CEO Genome Project
Manuel L. Gonzalez-Garay, Ph.D., Assistant Professor, The University of Texas Health Science Center at Houston
Replacing traditional methods for genetic testing of inheritable disorders with next-generation sequencing (NGS) will reduce the cost analysis and increase the information available for the patients. We organized a local event directed to educate healthy influential members of our community about genetics and NGS. During the presentation, we will explain our bioinformatics analysis and decisions to assure that our genetic diagnostics were accurate to detect carrier status and serious medical conditions on our volunteers.

2:05 Q&A/Discussion

2:15 Refreshment Break with Exhibit & Poster Viewing

The Funding Debate: What Insurers/Payors are Willing to Pay vs What Investors are Willing to Fund

3:00 Chairperson's Remarks
Steven Dickman, President & Owner, CBT Advisors

3:05 Good to Great: Patient Access and Broad Payer Coverage Can Be the Difference between a Good Product and One That Greatly Impacts Patients' Lives
Pam Baker, Senior Director, Market Access, CardioDx
Our test, Corus CAD, has achieved an important milestone in gaining Medicare coverage. This presentation will share learnings about the Medicare process, the commercial payer process, how they differ, and best practices for companies at various stages of development of genetic and genomic tests that are seeking payer coverage. Insights into how a company has successfully (and at times unsuccessfully) navigated the payer coverage process will be discussed.

3:25 Improving Preventive Care in Dentistry Utilizing Genetic Testing
Lynn Doucette-Stamm, Ph.D., Vice President, Development and Clinical Operations, Interleukin Genetics, Inc.
The value of preventive dental care is well established and an accepted part of healthcare. $108 billion is spent annually on dental care with 76% of dental procedures being dental examinations and cleanings. New evidence demonstrates that patients can be stratified for more effective and efficient preventive care based on specific factors including the results of our genetic test. With our partners, we have the ability to improve the delivery of preventive care to the benefit of dental patients and to allow dentists to play an active role in personalized healthcare.

3:45 The Investor Perspective
Gary J. Kurtzman, M.D., Managing Director, Healthcare, Safeguard
This presentation discusses criteria and metrics that are most important in an ideal investment.

4:05 How Will New Technologies Reach Patients in the Future?
Steve Gullans, Managing Director, Excel Venture Management
With regulatory and reimbursement challenges ahead, how will patients gain early access to new diagnostics and treatments? How will investors be able to make money?

4:25 Citizen Science and the Human Microbiome
Jessica Richman, CEO and Co-Founder, uBiome
The human microbiome is a rapidly growing field of scientific inquiry, to which uBiome is taking a unique approach: citizen science. We ran the largest successful citizen science crowdfunding campaign, and are now involving the public in asking and answering questions about the trillions of bacteria that live on and inside of us.

4:45 Q&A Discussion

5:15 Welcome Reception with Exhibit and Poster Viewing

6:30 Networking Party *(Sponsorship Opportunity Available)*

Day 2: Thursday, September 26

7:30 am Interactive Breakout Discussions Groups & Report Outs
Grab a cup of coffee and pastry and join your peers over stimulating discussions. These concurrent discussion groups provide an opportunity for all attendees, speakers, sponsors, and exhibitors to gather, participate in active idea sharing, and network with potential collaborators. Each discussion group will be limited to 12 participants and a moderator. Additionally, each group will take notes and report key findings in a large group share at the end. These notes will be available to attendees post-conference. See website for details.

Technology Company/Provider Spotlight: Current Trends and Visions

This session will feature a series of 20-30 minute presentations showcasing emerging and hot platform and software technologies relating to sequencing, data storage/management/analysis, modeling tools, website design, and more.

8:30 Chairperson's Remarks

8:35 LIVE: SAMPLE SEQUENCING ONSTAGE
John Boyce, President and CEO, GnuBIO
Unlike other sequencing instruments or "workflows" that require many days of sample preparation, or require large upfront expensive robotics, the GnuBIO system is the size of desktop computer and provides a fully-integrated solution. The researcher or clinician simply injects the patient genomic DNA into a cartridge with no need for target enrichment or library preparation, loads the cartridge into the instrument, hits the "Go" button, and in 3.5 hours receives the analyzed sequence of the gene panel including variant calls and quality scores. Both the small footprint of the system, as well as the price of the instrument $50,000 USD all in – no additional equipment or large robotics to purchase - will ensure rapid adoption of the GnuBIO solution. GnuBIO will do something that no other DNA analysis company has ever done before: 1) one person will carry the GnuBIO system on stage and plug it into the wall (our equivalent of the 3 day install required by most systems), and 2) a blinded genomic DNA sample will be injected into a cartridge for analysis. Results will be presented in 3.5 hours during the GnuBIO talk.

8:45 GenoSpace: Solving the Big Data Problem for 21st Century Genomic Medicine
Mick Correll, COO, Genospace
Today, genome sequencing is rapidly becoming a commodity so that the barrier to precision genomic medicine is no longer generating data, but securely storing it, analyzing it, and making it accessible and useful. At GenoSpace, we recognize that genomic data is fundamentally identifiable information and that addressing any of the challenges associated with its use starts with a secure infrastructure for storing it together with the associated clinical data. Beyond simple data storage, however, there are diverse groups—scientists, drug companies, health care organization, physicians, and the patients themselves—all of whom had different needs when accessing the same data. We'll describe why solving the problem of "Big" genomic data is so important and how we are assuring that the data is both appropriately used and useful.

9:05 Pharmacogenomic Testing: The Next Generation
Sponsored by GENEWIZ® Solid science. Superior service.
Guanghui Hu, Ph.D., Vice President, Translational Genomics, GENEWIZ, Inc.
GENEWIZ, Inc. developed two innovative and cost-effective next generation sequencing-based pharmacogenomic tests for personalized medicine: OncoGxOne™ and PGxOne™. OncoGxOne™ provides thorough coverage and assessment on cancer genes of interest with the ability to detect all types of genomic aberrations, including point mutations, Indels, rearrangements, and CNV. Similarly, PGxOne™ is capable of assaying all medically actionable germline genetic variations, including substitutions, Indels, and CNV, for drug selection and dosage in one single test

9:35 Enabling Market Access and Scalability for Consumer Genetics
Sponsored by DNA genotek
Ian Curry, President, DNA Genotek
When introducing new consumer genetics offerings it is imperative to consider all parts of the business model from sample collection to processing and validated results. Optimizing consumer experience, ensuring quality samples and proven performance on a range of assays is key to driving scalable and successful business models.

Main Conference

10:05 Coffee Break with Exhibit & Poster Viewing

10:55 Using a Patient's Genetic Information in the Real World

Michael Christman, Ph.D., President and CEO, Coriell Institute for Medical Research

Through the Coriell Personalized Medicine Collaborative (CPMC) research study we 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. Dr. Christman will speak to various challenges approached in a large cohort study, the CPMC, in which personal genomic information is reported to study participants and outcome measures are gathered to examine its utility. A partnership with the United States Air Force (USAF), which brings personalized medicine to 2,000 USAF Medical Service personnel, will also be discussed. Finally, lessons learned from the study and its 7,500 participants have been used to launch a commercial offering in partnership with IBM called Coriell Life Sciences. This new company aims to provide the needed technology infrastructure required to bring the era of genome-informed medicine to the clinic and will connect sequencing companies, data interpretation experts and physicians.

11:25 Sponsored Presentation *(Opportunities Available)*

11:55 Establishing the First CLIA Whole-Genome Sequencing Laboratory: Best Practices and Lessons Learned

Sponsored by illumina

Tina Hambuch, Ph.D., Associate Director, Illumina, Inc.

The barriers to accurate human genome re-sequencing have largely been surmounted, enabling considerations of routine clinical applications. Significant challenges remain for interpretation of whole genome data for diagnostic or prognostic purposes. We established a clinical service for whole genome sequencing, and describe here the challenges of implementing clinical interpretation for it. The outcome makes a compelling case for the need for a clinical grade database based on standards established by the medical and genetics communities.

12:25 pm True Clinical Desktop DNA Sequencing: Sample In, Answer Out

John Boyce, President and CEO, GnuBIO

12:55 pm Luncheon Presentation *(Sponsorship Opportunity Available)* **or Lunch on Your Own**

Communicating Genetics and Science to Everyone

1:55 Chairperson's Remarks

Peter S. Miller, COO, Genomic Healthcare Strategies

2:00 A Communications Overview of Consumer Genetics

Peter S. Miller, COO, Genomic Healthcare Strategies

This talk will provide an overview of the various forms of communication which will emerge as the consumer genetics market grows and matures. As consumer genetics become commonplace, a web of communications will connect consumers, patients, physicians, businesses, medical records, payers, and labs. Organizations seeking to have a strong presence in consumer genetics will need to build their communications capabilities.

2:20 Merging Art, Science, and Process: Key Strategies for Designing Great Experiences

Michael Hawley, Chief Design Officer, Mad Pow

During this fast-paced session, we'll discuss several core principles of the science of human factors and the art of design that lead to successful interactions. Using examples from several consumer genetics applications and services, we'll evaluate current design trends in the genetics marketplace and demonstrate how well designed products take advantage of successful design strategies.

2:40 Using Data Visualization Tools and Strategies to Help Consumers Understand Their Genetic Results

Kenneth Chahine, Ph.D., J.D., Senior Vice President and General Manager, DNA, ancestry.com

An important, but often overlooked, challenge in consumer genetics is the design of engaging and informative data visualization strategies that help consumers understand and fully appreciate the results of their genetic results. At AncestryDNA, we have considered this problem in the context of admixture prediction based on autosomal SNP testing. We will present our findings based on the conviction that the principles we have applied in our iterative development, testing and refinement of user experiences can also extend to other aspects of consumer genetics.

3:00 Hacking Health

Juhan Sonin, Creative Director, Involution Studios, MIT

Hacking Health will look at the design, data, and policy challenges, as well as the scary new ideas being invented to solve them, that make tackling the healthcare problem daunting and exciting. Healthcare is deadly. As patients and clinicians, we have long passed the threshold of comprehension when it comes to the amount of data and variables we are given to make life-critical decisions. The system needs to be redesigned and re-imagined. True change comes from establishing a vision for healthcare that will modernize how doctors and patients interact with their health data. In "Design is Medicine," design provocateur Juhan Sonin will show how linear-thinking human beings can interpret non-linear, unbalanced health metrics, and how healthcare can be saved through coordination, presentation and design.

3:20 Sponsored Presentation *(Opportunity Available)*

3:40 Q&A Discussion

3:50 Refreshment Break with Exhibit & Poster Viewing

Consumer Testing – Points and Counterpoints

4:30 Chairperson's Remarks

4:35 Direct-to-Consumer Pharmacogenomic Testing: Helpful or Harmful?

Cinnamon S. Bloss, Ph.D., Director, Social Sciences & Bioethics, Assistant Professor, Scripps Translational Science Institute

The availability of direct-to-consumer (DTC) genomic testing has generated controversy in the scientific and clinical communities. While a small number of studies have evaluated the behavioral and psychological impacts of DTC genomic testing for complex disease risk on consumers, studies that assess the impact of pharmacogenomic (PGx) risk profiling are lacking. This presentation will focus on data from the Scripps Genomic Health Initiative (SGHI), a large longitudinal cohort study designed to assess the impact of DTC genomic testing on consumer behavior. DTC PGx risk profiling among SGHI participants was associated with statistically significant increases in self-reported healthcare utilization and did not result in any adverse changes in psychological health or follow-up test-related distress.

4:55 Pros and Cons of DTC Genetic Testing

Ellen T. Matloff, MS, Research Scientist, Department of Genetics and Director, Cancer Genetic Counseling, Yale Cancer Center

Genetic testing companies are now offering direct-to-consumer (DTC) tests via kits sold on the Internet, including tests for risks of several cancers. However, such tests are typically based on single nucleotide polymorphism (SNP) panels whose accuracy has not been validated. This talk explores the pros and cons of DTC genetic testing for both consumers and physicians, illustrated with case vignettes.

Main Conference

5:15 N=1 Human Study in Clinical Neurosciences: Genomic Guided Medicine and Deep Brain Stimulation

Gholson Lyon, M.D., Ph.D., Assistant Professor of Human Genetics, Cold Spring Harbor Laboratory; Research Scientist, Utah Foundation for Biomedical Research

There is an extraordinarily rich clinical literature in clinical psychology, psychiatry and neurology of N=1 studies. This presentation discusses the detailed phenotypic characterization, clinical-grade whole genome sequencing (WGS), and two-year outcome of one man with severe obsessive-compulsive disorder successfully treated with deep brain stimulation (DBS). To my knowledge, this is the first N=1 human study in the clinical neurosciences including 1) detailed neuropsychiatric phenotyping, 2) clinical-grade WGS with management of genetic results for a person with severe mental illness, and 3) individualized treatment with deep brain stimulation for his OCD. His WGS results and positive outcome with DBS for OCD is one example of individualized medicine in neuropsychiatry, including genomics-guided preventive efforts and brain-implantable devices. We have archived and offered the genome data to him, so that he and others can manage and re-analyze his genome for years to come. This presentation discusses the implications of this for consumer genetics and the future of medicine.

5:35 Genome Hacking

Yaniv Erlich, Ph.D., Principal Investigator and Whitehead Fellow, Whitehead Institute for Biomedical Research

Sharing sequencing datasets without identifiers has become a common practice in genomics. We recently showed that some datasets can be fully re-identified by using entirely free, publicly accessible Internet resources. This talk will present quantitative analysis about the probability of identifying U.S. individuals by this technique on hundreds of genetic datasets. In addition, this talk will demonstrate the power of our approach by tracing back the identities of multiple whole genome datasets in public sequencing repositories.

5:55 Q&A Discussion

6:05 Featured Presentation: Impact of Patent Law on the Future of Genetic Testing

Antoinette F. Konski, J.D., Partner, Foley & Lardner LLP

The U.S. Supreme Court has changed the landscape of IP protection of genes and their use diagnostically. This presentation will discuss the current understanding of IP protection of human genes and their use in medical testing.

6:35 - 8:30 Networking Party Hosted by GnuBio

Day 3: Friday, September 27

7:30 am Interactive Breakout Discussions Groups & Report Outs

Grab a cup of coffee and pastry and join your peers over stimulating discussions. These concurrent discussion groups provide an opportunity for all attendees, speakers, sponsors, and exhibitors to gather, participate in active idea sharing, and network with potential collaborators. Each discussion group will be limited to 12 participants and a moderator. Additionally, each group will take notes and report key findings in a large group share at the end. These notes will be available to attendees post-conference. See website for details.

Digital Consumer Healthcare: Health Tracking Applications, Demos & Case Studies

8:30 Chairperson's Remarks

8:35 Oversight = Opportunity: Regulation, Innovation and the Power of Mobile Health Apps

Martin Mendiola, M.D., MPH, Director, Clinical Program Development, Happtique

With over 40,000 health care apps in the app marketplace and little barrier to entry, patients and health care professionals have little information about app security, credibility, and functionality. Happtique's Health App Certification Program was developed to address those issues. With the right kind of industry oversight, apps have the power to not only change the way patients manage their health, but transform patient-centered care across the entire industry.

8:55 DTC 2.0: Genomics in a Digital Health World

Anish Sebastian, Co-Founder and CEO, 1eq

Julia Oh, CSO, 1eq

1eq has created a consumer digital health platform that integrates genetics and lifestyle information to produce a clear health picture with actionable items for its user to live a better and healthier life. Case studies will be presented to show how this platform has changed user behavior. The 1eq App offers a unique DTC technology to the masses by making genomics a piece of the overall health puzzle.

9:15 Innovative Solutions for Effective Disease Management

Georgia Mitsi, MSc, Ph.D., MBA, Founder and CEO, Apptomics LLC

Apptomics is a digital health company developing innovative solutions for neurodegenerative diseases. We will describe a mobile application that allows daily data capture (disease features, severity, medication log, etc.) in combination with a wearable sensor system that objectively measures motor symptoms. Patients stay involved with their health, caregivers can offer their help in a meaningful way, and doctors can make more informed decisions that lead to more effective disease management. Attendees will understand how the new technologies can help Parkinson's patients understand the challenges associated with new technologies in healthcare.

9:35 Quantified Self 2.0

Ayub Khattak, CEO, ruubix

Tracking our real-time hormone levels (testosterone, Vitamin D), inflammation index (CRP), cholesterol and other important molecular indicators of body-state are an important addition to tracking our heart rate, activity, and sleep, which are already well covered by wearable monitors. We are developing a mobile-based platform for regularly measuring several of these biomarkers which are of interest to the general population for meeting fitness, weight, mood, and wellness goals. The complete picture of health and wellness can be achieved by combining and analyzing data from your genetic disposition (i.e. 23andMe), your diet and activity (i.e. FitBit, Jawbone) and your body's real-time molecular response at the hormonal and protein level to how you eat, sleep, and exercise.

9:55 Sponsored Presentation *(Opportunity Available)*

10:15 Coffee Break, Last Chance for Exhibit & Poster Viewing

Consumer Landscape: Current Opportunities and the Big Picture

10:55 Be The Match®, The National Marrow Donor Program's Role in Consumer Genetics

Eric P. Williams, Ph.D., Senior Bioinformatics Scientist, National Marrow Donor Program

Established in 1986, The National Marrow Donor Program (NMDP) operates the Be The Match® registry and oversees operation of the C.W. Bill Young Program mandated by Congress. For most of the 10.9 million volunteer donors and 185,000 cords listed on the registry, typing is done at the DNA level and covers five genes in the Major Histocompatibility Complex (MHC) of chromosome 6. This region governs the immune system, pharmacogenomic and disease interactions and can be used to determine ancestral origin. The highly polymorphic MHC complicates the process of determining reliable genotype results for most DNA sequencing technologies including Next-generation Sequencing. There are numerous synergistic relationships that would aid NMDP efforts to increase their registry size and offer genetic population, typing results interpretation, reference sequence, and geospatial orientation help to consumer

genetics providers interested in enhancing typing results for the MHC. The NMDP has expertise that would help consumer genetics providers determine results for an area of the genome that is notoriously complicated and hard to interpret. We will present an understanding of NMDP's mission and areas of expertise, areas for collaboration, and how we could be helped by providers.

11:15 Alice in Genomeland: Lessons from A Reluctant Biotech Entrepreneur

Anne Morriss, Founder and CEO, Genepeeks

From diagnosis to fundraising to product launch, this talk captures the key lessons from one mother's experience turning her son's inheritance of a rare genetic disorder – MCAD Deficiency -- into the inspiration for a consumer genetics company. The company, Genepeeks, predicts the genetic profile of a child pre-conception by simulating the genetic reduction and recombination that occur naturally during reproduction, digitally weaving together the DNA of prospective parents.

11:35 Sponsored Presentation *(Opportunity Available)*

11:55 Consumer Genomics: From Niche to Mainstream

Spencer Wells, Ph.D., Explorer-in-Residence and Director, The Genographic Project, National Geographic Society

The world of consumer genomics has transformed over the past decade. What was once an expensive, niche hobby limited to a small number of genealogists has exploded. 2013 saw the millionth person tested, and around half of these have done so through National Geographic's Genographic Project. I will discuss Genographic as a model for consumer engagement, and speculate on longer-term trends in the industry as testing goes mainstream.

12:40 pm Q&A/Discussion/Closing Remarks

1:00 Close of Conference

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