

## Executive Summary

### Personalized Health Care Initiative Workshop: “Understanding the Needs of Consumers in the Use of Genome-based Health Information Services”

July 7, 2008  
Washington, D.C.

Transcript: [have the document]

Webcast: [have the video files]

### **Background**

In March 2007, the Department of Health and Human Services (HHS) launched the Personalized Health Care Initiative to advance the integration of gene-based technologies into health care delivery. The Initiative is aimed at helping to achieve a new paradigm for health care in which interventions may be better tailored to each individual based on his or her unique gene-based characteristics. This vision stems from recent advances in science and technology and emphasizes the potential for prevention and early detection of disease.

The completion of the Human Genome Project marked a huge scientific accomplishment and initiated a burst in scientific discovery of the genetic underpinning of diseases. The development of faster and less expensive genotyping technologies has helped propel this burst of discovery. Since early 2005, more than 100 genetic associations have been identified for 40 common diseases utilizing these technologies.

Efforts are underway in the private and academic sectors to find new ways to apply these discoveries for the benefit of consumers. Recently, several new services have been launched, aimed specifically at the consumer market. These services offer access to one's own genomic information and provide health-related and other information based on genotype, family history, and other medical features. These new services have occasioned considerable press and public attention. They come at a time when the medical community is undertaking a more engaged role in incorporating genomic resources into health care. Companies offering these services maintain that this information represents a new phase in empowering, engaging, and educating consumers in their health and medical decision-making. Meanwhile, others have questioned the value of these services for consumers, especially at this early stage of knowledge development. Concerns include unsubstantiated claims and consumers being provided unwarranted false sense of security, resulting in consumers' avoidance of healthy behavior based on presumed lack of risk.

Through the HHS Office of the Assistant Secretary for Planning and Evaluation (ASPE), the Personalized Health Care Initiative sponsored a workshop in Washington, D.C., on July 7, 2008 entitled “Understanding the Needs of Consumers in the Use of Genome-based Health Information Services.” This workshop was aimed at enabling various stakeholders to share differing perspectives and discuss alternatives for helping consumers make informed use of genome-based health information, both now and in the future.

### **Opening Remarks: Meeting Objective and Overview**

Richard Campanelli, J.D., counselor to the Secretary of HHS for Science and Public Health, welcomed the participants to the meeting and described the vision for personalized health care. Mr. Campanelli described the convergence of genomic science, technological advancement, and consumer interest. This convergence has fundamental importance for personalized health care, because of new opportunities for consumer engagement and for prevention that it presents. He encouraged the participants to consider the opportunities, necessary cautions, and the current steps that can be done to help achieve the best possible outcomes as personalized health care capabilities and opportunities emerge. The objective of the workshop is to enhance understanding and provide an early opportunity for public dialogue among interested groups as this new area of science and technology develops and interfaces with consumers.

### **Opening Remarks: Empowered Medical Decision Making**

Eric J. Topol, M.D., Director of Scripps Translational Science Institute, and Dean of Scripps School of Medicine, described how medical decision making can be empowered by genomic information. Examples included how genomic information may redefine disease and aid in diagnosis of macular degeneration, myocardial infarction, abdominal aortic aneurysm, intracranial aneurysm, type 2 diabetes, breast cancer, and prostate cancer.

In addition, Dr. Topol described the current companies that provide consumers access to their genomic information, the potential for consumer empowerment, and the medical community’s reaction to this emerging area. He suggested that consumers armed with this information may prod clinicians to become more informed about genomic medicine. He sees a future where clinicians and consumers will experience routine personalized medicine based on genomic information.

### **Opening Background: Consumer Interest in Health and Genomic Information**

Steve Bodhaine, M.B.A., Group President of Yankelovich, Inc., the market research based consulting firm, described consumer interest in health and genomic information, drawn from long-term market research into consumer trends and lifestyle as well as recent individual interviews. In trying to understand consumer perspectives, Yankelovich has identified six general types of consumers. These groups vary in their motivations for

health. Bodhaine emphasized that consumers view their lives holistically, and people define terms such as *health*, *healthcare*, *wellness*, and *well-being* very differently.

In its qualitative research work in the spring of 2008 to understand consumer interest in genomic information, Yankelovich found that consumers demonstrated familiarity with genetic testing, but they are not sure what it involves, how the information is used, what the results would look like, or how much it would cost. Consumers are largely unaware of genetic testing information companies that provide services via the Internet. The healthcare professional is desired – and assumed – to be involved in the information gathering, testing, and analysis processes. Most consumers say they would consider genetic testing, especially for actionable conditions. Consumers wanted a “reputable organization” to create an industry standard to help consumers identify which companies were trustworthy, though they mentioned that the Internet would be a likely source to turn for obtaining information. Privacy of test results was assumed by consumers, and greater concern was expressed about accuracy of tests.

### **Panel 1: What is the consumer interest in genome-based health information?**

Panel 1 was moderated by Esther Dyson (EDventure Holdings) and was comprised of Linda Avey (Co-founder of 23andMe, Inc.); Rebecca Fisher, MLIS (Patient Advocate); and Matthew Holt (Co-founder Health 2.0 Conference) who addressed the following questions:

- What drives the consumer’s interest in health information for predicting health outcomes?
- What are key factors for motivating interest in genetic testing?
- For what information are consumers ready?

The panel identified gaps in consumer knowledge and concluded that individual need and curiosity may motivate interest in genetic testing.

#### Gap in knowledge

Ms. Fisher cautioned about providing research-grade data to consumers. She shared her experience as a BRCA1 breast cancer survivor, which was diagnosed at age 31. In general, she noted a gap between what consumers/clinicians presently know and what they may need to know.

The panel discussed consumers’ ability to acquire information and methods for providing information to consumers. All acknowledge that genetic information is complex, but it was compared to baseball statistics – as people become familiar with the vernacular and the complexity, they acquire the ability to use and understand even complex information.

Ms. Avey suggested that the internet is an excellent medium for providing information in layers – according to the reader’s interest level. By providing layers of details, consumers can access the level of detail they desire.

#### Motivation for obtaining personal health information

Mr. Holt suggested that a motivation for seeking health information is often the onset of a condition or disease. This motivates the consumer to seek more information about the disease and possible interventions. Recently, online communities have emerged where patients share information about their conditions and treatments to educate and help others who have the same disease.

One motivation for seeking genetic information about one's self seems to be curiosity. 23andMe responded to customer requests to provide more genomic information by reporting less scientifically validated associations. To convey the variability in scientific validation, 23andMe created a star system to rank the scientific basis for a particular trait or disease.

## **Panel 2: Is the testing process reliable, and is the information's privacy maintained?**

Reed Tuckson, MD (Executive Vice President and Chief of Medical Affairs, UnitedHealth Group) moderated a panel comprised of Jeffrey Gulcher, MD, Ph.D. (Chief Scientific Officer, deCODE genetics); Deven McGraw, JD, MPH (Director, Health Privacy Project, Center for Democracy and Technology); and Ryan Phelan (Founder and CEO, DNA Direct). The second panel addressed the following questions:

- How are risk assessments determined?
- What are potential ways to inform consumers that quality control standards for testing are being met?
- Will (should) the genomic data be utilized for public health benefit or other population-based research in the future?
- Are there cautions that the consumer should consider when sharing their genomic information with others, such as family members, members of social network, or clinicians?
- What protections are currently or should be employed to protect the security and privacy of consumer genomic information?

### Type of genetic testing and test accuracy

The panel discussed the type of genetic testing, the information that is currently provided to consumers, and the level of interaction from the health professional. Diagnostic testing for genetic diseases, such as Huntington's disease, is not provided by any of the consumer-oriented companies because if one has the mutations associated with Huntington's disease, one will develop the disease, and the companies agree that this type of information requires physician oversight. DNADirect provides access to predictive testing for serious disease (to determine risk for disease onset) and for genetic screening for carrier and drug response, conducted with physician oversight and phone/web-based genetic counseling. The genome-wide scanning (performed by 23andMe, deCODEme, and Navigenics) is generally viewed as a different type of test and the type of support services available may be dependent on the situation or condition.

Regardless of the type of genetic testing, establishing the analytic validity (the testing is done accurately) is readily accomplished. Adhering to the standards set by Clinical Laboratory Improvement Amendments of 1988 (CLIA) was understood to describe analytic validity needed for this testing. However, communicating the meaning of CLIA-certification to consumers may not be as straightforward demonstrating compliance.

### Risk Factors

Dr. Gulcher compared genome scanning to medical diagnostic testing. The interpretations derived from genome-wide scans provide risk factors – not guarantees for disease onset, as in Huntington’s disease. For the most part, the information from the SNPs are of modest effect, but Dr. Gulcher suggested that these risk factors can be combined with other known risk factors of the same magnitude (such as environment, age, weight, family history) to inform medical decision-making. In some cases, prostate cancer, for example, may be more suited for early diagnosis by genetic testing than the current standard of using family history to identify candidates for early screening as 95% of prostate cancer patients have no family history of prostate cancer. There is some discrepancy among the consumer-oriented companies performing genome-wide scans for converting the odds ratios typically reported in scientific literature to risk ratios.

### Privacy and Security

Ms. McGraw discussed that consumers need to have a much better understanding of the appropriate uses of their information. Privacy aspects go beyond protecting the data or information from theft or breach. When consumers seek a genetic test, they should have a complete understanding of what may be done with the information. She said that current legal protections are incomplete.

### **Panel 3: What is currently useful to consumers, and what can they expect in the future?**

The Honorable Nancy Johnson (former Congresswoman and currently Senior Public Policy Advisor at Baker Donelson) moderated a panel comprised of Mari Baker (President and CEO, Navigenics); Katherine Johansen, Ph.D. (Senior Scientist, American Medical Association Program in Genetics and Molecular Medicine); Ronni Sandroff, MA (Director/Editor, Health and Family Information, Consumer Reports); and Angela Trepanier, MS, CGC (President, National Society of Genetic Counselors and Assistant Professor, Wayne State University). The panelists addressed the following questions:

- How can genomic information and explanatory resources deepen health literacy and support consumer empowerment for prevention?
- What checklist of resources or information should consumers have to assign value to genome-based health information services?
- How valuable is the information currently available relative to other health information, such as family history, blood pressure, BMI etc.?
- What can/should consumers expect in the future?

The panel described some challenges for integrating genetics to practice and considered information that consumers might need.

### Challenges to ingrate genetics to medical practice

The current health care system is based on treating illness and may be less equipped for prevention. However, some panelists suggested that improving the scientific basis for prevention and engaging patients more in their own care may help with this transition. Genetic test information may motivate a consumer to engage in healthy lifestyle choices or may spur a conversation with a health care professional about potential interventions.

Ms. Trepanier emphasized that this type of personalized medicine requires more than just knowing the consumer's genetic disease risks, but requires the health care professional to adapt the method for presenting information to each consumer. Each consumer brings unique concerns, preference for absorbing information, and motivation for making changes.

In considering information that is currently useful for informing care decisions, Ms. Baker reiterated that current clinical decisions are made by combining risk factors from family history, environmental factors, lifestyle choices, age, and weight. In some areas, genetic testing may provide an additional risk factor that can be incorporated into a clinical decision.

Dr. Johansen mentioned that the American Medical Association (AMA) had recently revised its policy on direct-to-consumer genetic testing and now recommends that consumers who elect to use such services engage with their clinician to interpret genomic information. She said clinical decisions should be made in consultation with health care professionals such as nurses, nurse practitioners, pharmacists, physicians, and physician assistants although these health care professionals may not be as prepared as genetic counselors to advise on genetic information. Current efforts by AMA, Medscape, Mayo Clinic, and others are providing continuing medical education for genetics and helping revise training curricula. In the future more consumers will have genetic information as part of their health record and this underlines the need for clinicians to have genetics training.

Additional concerns for the future for this type of information include creating a new type of health disparity (a genetic divide) where predictive, genetic information might be available primarily for the affluent.

### Consumer information needs and concerns

Ms. Johnson cautioned against over-selling the promise of genomic medicine, as it is still developing. She suggested that the best protection against that danger is for the private and public sectors to work together to achieve the transparency, openness, and directness

necessary to gain and maintain public trust. Panels 2 and 3 agreed that consumer genomics companies need to do the following:

- Present information on the test capabilities and limitations
- Communicate the value of products
  - What will this test actually do for me?
  - Will this test help me make a better healthcare decision?
  - Will these tests give me peace of mind?
- Explain concepts of relative risk
- Identify fraudulent or misleading claims
- Describe the method for combining risk factors
- Provide summary document structured for the clinician
- Ensure the privacy and security of results
  - Who has the information?
  - Is the information linked to identifiable information?
- Identify insurance reimbursement

#### Development of industry standard

It was announced that consumer oriented companies will be working together to define best practices or standards for the consumer genomics industry. This effort will be facilitated by the Personalized Medicine Coalition, which represents a broad spectrum of academic, industrial, patients, providers, and payer communities that seek to advance the understanding and adoption of personalized medicine concepts and products for the benefit of patients.

Participants agreed that this effort should address the following issues: communicating analytic validity to consumers; methods for combining risk factors; techniques for presenting risk factors to consumers and health care professionals; discussion of the transition point from a test that provides information to one that has clear clinical implications and whether the latter type may benefit from involvement by a clinician (genetic counselor, nurse practitioner, physician assistant, or physician); and privacy and security standards.

In addition, Ms. Phelan described a concept to collaborate with the industry, non-profit and academic institutions, and other experts to help consumers identify whether or not a test is going to be useful, responsible, and relevant to them. This evaluation may contain scientific validity, predictive value, clinical utility (identifying the outcomes associated with specific test results), and personal utility of the test (value of the information for the recipient).

#### **Looking to the Future of Consumer Services**

Michael Cowan, MD, chief medical officer of BearingPoint and former Surgeon General of the Navy, summarized the meeting. The workshop dialogue provided clear evidence of the growing pains for a science moving very fast; emerging from a science into a young industry; and trying to figure out how to handle the risk, the science, the motivations, the

markets, and the trust to develop into a successful system to benefit consumers. Dr. Cowan identified a few points of convergence: the potential of genomics to inform medical decision making and the need to communicate risk to consumers and clinicians. He also enumerated areas of current divergence: when consumers should have access to information; when and how to engage clinicians; what are the appropriate levels of oversight and regulation; and there are divided views on privacy, the reliability, and integrity of information. In addition, Dr. Cowan encouraged the participants to consider the ethics as the science and technology develop instead of waiting for problems to emerge after products and services are marketed.