



PRESS RELEASE

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Researchers Shed Light on Implications and Impact of Using Direct-to-Consumer and Clinical Genetic Testing in Disease Risk Assessment

Scientists Present Latest Research Findings on the Public's Understanding, Use, and Attitudes Toward Personalized Genetic Testing at ASHG 60th Annual Meeting

BETHESDA, MD – October 20, 2010 – Thousands of the world's top scientists and clinicians in the human genetics field will convene to present their latest research findings at the [American Society of Human Genetics 60th Annual Meeting](#), which will be held November 2-6, 2010, in Washington, D.C.

A number of scientific presentations at this year's meeting will provide information about important new research findings that advance and expand our current understanding of the issues and implications involved in direct-to-consumer and clinical genetic testing. ASHG will host a press briefing at the 2010 Annual Meeting to highlight three presentations of interest on this topic.

ASHG invites members of the media to attend this press briefing event titled, "*Researchers Shed Light on Implications and Impact of Using Direct-to-Consumer and Clinical Genetic Testing in Disease Risk Assessment*," which will be held on Friday, Nov. 5, 2010, from 12:00-1:00 p.m. in the ASHG Press Briefing Room at the Walter E. Washington Convention Center (Room #101). **[To view the online webcast recording of this press briefing session after the event has ended, please go to: http://www.ashg.org/genetictestingissues/](http://www.ashg.org/genetictestingissues/)**

Session moderator and President-Elect of ASHG, [Lynn Jorde, PhD](#), will open with a brief introduction to the topic area and an overview of [ASHG's position statements on direct-to-consumer \(DTC\) genetic testing](#) use and oversight in the U.S., and on the recently proposed [retail sale of genetic test kits](#) to the public in drugstores across the nation. After Dr. Jorde's introduction, the other three speakers will present their latest research findings from the following abstracts on DTC and clinical genetic testing studies:

- **David Kaufman, PhD** – ["*Direct from Consumers: A Survey of 1,048 Customers of Three Direct-to-Consumer Personal Genetic Testing Companies about Motivations, Attitudes, and Responses to Testing*"](#)
- **Barbara Bernhardt, MS, CGC** – ["*Public Understanding of and Reactions to Personalized Genetic Risk Information: Results from the Coriell Personalized Medicine Collaborative*"](#)
- **Andy Faucett, MS, CGC** – ["*The Use of Current Genetic Testing Oversight to Select the Best Test for Each Patient*"](#)

Brief summaries of the content covered in these three presentations are included in the section below:

Direct from Consumers: A Survey of 1,048 Customers of Three Personal Genetic Testing Companies about Motivations, Attitudes, and Responses to Testing

For several years, companies have been selling genetic tests which claim to predict a person's risk of developing disease. These tests currently cover up to 174 different conditions. Although doctors' offices have offered genetic tests for a number of these diseases for many years, these companies have made their mark by selling genetic testing services directly to consumers (or DTC), and some companies even provide tests that have not yet been adopted for routine medical practice. Concerns about the limitations, potential harms, and lack of clear benefits of these DTC tests abound. However, very little data has been published about who is buying these tests and why, whether the tests meet customers' expectations, whether customers understand the information that is reported in their personal genetic profile, and how they are using the resulting information.

To answer some of these important questions, a group of researchers led by [David Kaufman, PhD](#), Director of Research and Statistics for the Genetics and Public Policy Center at Johns Hopkins University, conducted an online survey of 1,048 customers who ordered DTC genetic testing from one of three different companies – 23andMe, deCODEme, and Navigenics – to assess their motivations and experiences using these services. Random samples of U.S. customers who received DTC results between June 2009 and March 2010 were invited to participate in the study. Survey question topics included reasons for testing, attitudes about test results, and health behaviors. Respondents were also asked to interpret two genetic test results shown using graphics and text provided by the DTC companies.

Dr. Kaufman's team found that early adopters of personal genomic DTC tests – who on average tended to be highly educated and have higher incomes – indicated that they were generally satisfied with their DTC genetic testing services. The top reasons that they cited for purchasing the tests were to satisfy their curiosity (94% said this was somewhat or very important), to assess their elevated risks of genetic diseases (91%), and to learn more about their ancestry (90% among the customers of 23andMe and deCODEme, which both offer ancestry testing). One in three participants said that they were interested in pursuing testing because a first-degree relative was affected with a tested genetic health condition, and 42% said they were interested in learning their risk of one or more specific conditions.

Overall, 77% of participants pursued testing to help improve their health, 58% said they learned new information that would help improve their health, and 9% felt they could not change their health risks after receiving their test results. Three in 10 study participants indicated that they had shared their results with a doctor or had gotten a follow-up test, and another 18% said that they intended to do so at a future date. As a result of testing, 34% of the participants said they were being more careful about their diet, 15% had changed their medication or dietary supplement regimens, and 14% were exercising more. Long-term follow-up of DTC test users is needed to evaluate the impact of DTC testing on healthcare usage and health behavior change.

The researchers also asked participants questions about how they viewed and interpreted the data provided in their personal genetic test result reports. Although 88% of customers agreed that their risk results were easy to understand, 38% of the study participants indicated that the information they received from the DTC companies was too vague. Furthermore, when customers were shown two examples of risk results provided by the companies, between 4% and 7% misinterpreted them. The participants were also twice as likely to misinterpret a result showing a gene that protected a person from disease, as compared to a result showing a gene that increased disease risk.

The study participants answered questions about their thoughts and opinions on federal regulation and oversight of DTC genetic tests. Although 66% of the participants felt that these tests should be available without government oversight, more than 70% indicated that an organization such as the Federal Trade Commission (FTC) or Consumer Reports should monitor companies' claims for scientific accuracy. "Our research results indicate that consumers who are at the front of the line to buy direct-to-consumer genetic testing services were motivated by curiosity about their genes, though many also hoped to find out their disease risk and improve their health with the results," said Dr. Kaufman.

“We observed that most of the DTC customers who participated in this study were generally satisfied with the personal genetic testing services that were provided, and more than half of the participants indicated that their test results helped them learn something new that could improve their health,” Kaufman noted. “However, many of the study participants found the information from their genetic test reports to be vague, and one in 12 was not able to correctly interpret the sample results that we showed them, suggesting that there is room to improve the clarity of the information provided to customers in their personal genetic test result reports and the way the information is being delivered to them.”

Public Understanding of and Reactions to Personalized Genetic Risk Information: Results from the Coriell Personalized Medicine Collaborative (CPMC)

Currently, little is known about how people interpret and react to personalized genomic risk information for common complex conditions. To address this knowledge gap, a group of researchers led by [Barbara Bernhardt, MS, CGC](#), Clinical Professor of Medicine and Co-Director of the Penn Center for the Integration of Genetic Health Care Technologies at the Hospital of the University of Pennsylvania, conducted interviews with individuals who received personalized genetic risk results for seven common health conditions through the [Coriell Personalized Medicine Collaborative \(CPMC\)](#), a research project examining the clinical utility of personalized genomic risk information. The interviews included questions about participants’ understanding of their disease risk and the actions they took based on their results.

From the 60 interview transcripts analyzed in this study, the researchers found that most people appeared to have a good understanding of their results, although some had difficulty interpreting relative risk values. Participants tended to interpret their risks based on genetic test results combined with their own family and medical history. Although some participants were surprised to learn about an increased genetic risk for a disorder that was not in their family, no participants reported being overly concerned about their risks.

About one third of the study participants acted on the information provided in their report by changing their lifestyle and/or health habits, but most of the participants reported only slightly modifying healthy behaviors that they were already practicing (including eating a healthier diet, exercising, taking vitamins, or using sunscreen). Only a few participants said they initiated a new risk-reduction behavior. Those who reported no behavior change either felt they were already doing everything they could to reduce their risk, or did not feel their level of risk was high enough to warrant any changes.

Nearly half of the study participants reported that they shared their results with a physician, and most of the others said they were planning to share the results with their doctor at a future date. The participants shared their results based on the belief that doctors would be interested in knowing this information and it would aid them in providing appropriate recommendations to reduce disease risk. Most of the participants who shared their results believed that their doctor had a good understanding of the results. However, about a quarter of the study participants indicated that their doctor did not understand the results, or did not know what to do with them. Recommendations made by doctors (e.g., lose weight, test blood lipid levels, lower dietary fat) followed standard population recommendations.

“Through our interviews with individuals receiving information about their genetic risk for common complex health conditions, we found that most of the study participants had a good understanding of personalized test results predicting their disease risk,” said Bernhardt. “We also found that these individuals were neither unduly worried nor falsely reassured by their results, and that they are likely to take steps to reduce risk that are aligned with the level of their disease risk.”

Bernhardt noted that, “Further research is needed to determine if these positive outcomes are generalizable to broader populations, and whether they can be achieved when testing is not accompanied by the type of extensive pre- and post-test education made available through the Coriell project.”

The Use of Current Genetic Testing Oversight to Select the Best Test for Each Patient

In the U.S., most genetic tests are developed by laboratory experts; however, many clinicians and the public remain unaware that genetic tests are *not* independently evaluated to ensure their efficacy and accuracy. Recently, several national groups – including the [American Society of Human Genetics](#) – have called for closer scrutiny of genetic testing. For example, the CDC has issued guidelines for laboratories,

the FDA is considering reviewing laboratory-developed tests, and the [NIH is developing a Genetic Test Registry](#). These efforts on the part of federal agencies and professional organizations to develop independent oversight for genetic tests must ensure that the tests are effective, without stifling innovation or raising the cost of genetic testing. Taking these issues into consideration, given the current lack of independent oversight and federal regulation of clinical and DTC genetic tests, it is important that both healthcare providers and consumers evaluate each test in light of its specific use, its reliability and accuracy, and whether or not the test results will provide useful information.

According to [Andrew Faucett, MS, CGC](#), Director of the Genomics and Public Health Program at Emory University School of Medicine, although healthcare providers evaluate other types of medical tests on a daily basis, many clinicians are not experienced in evaluating genetic tests. However, it is important for healthcare providers to carefully review clinical genetic tests to ensure that their patients are offered tests that will provide the most accurate and appropriate information at a reasonable cost. Faucett suggests that the most important questions for practitioners to ask when evaluating genetic tests are:

- What is the test analyzing, what is it *not* analyzing, and what will the information learned from the test be able to tell you?
- Will the test be able to provide information that can be used to guide patient treatment or help inform clinical care decisions?
- Is the test looking for a single mutation or all known mutations for a gene?
- Has the test been implemented and tested in the correct populations for the patient?

For consumers interested in using DTC testing services, Faucett indicates that it is important for them to review tests critically and/or consult with an expert in genetic testing. He suggests that consumers considering DTC genetic testing should ask the following questions beforehand:

- Will the information that I gain from this genetic test help answer my questions about inherited disease risks, ancestry, etc.?
- Will the information be useful in helping to improve my health or make better, more informed decisions about my medical care?
- Is there solid evidence to back up the clinical utility and validity of this particular genetic test?

“Most U.S. laboratories performing genetic testing do an excellent job of analysis – however, that said, sometimes the test results may not provide the appropriate information to address the questions that the healthcare providers or patients are asking,” said Faucett.

“It is important for both healthcare providers ordering clinical genetic tests for their patients and individuals who are considering purchasing direct-to-consumer genetic testing services to evaluate these tests carefully before making decisions,” Faucett notes. “Consumers, in particular, must be wary of reading too much into their test results, and they may even want to consult with a certified genetics professional to make sure they’re receiving a test that will provide appropriate information that could help them improve their health and assist doctors in making better, more informed medical care decisions.”

ABOUT THE AMERICAN SOCIETY OF HUMAN GENETICS

Founded in 1948, the American Society of Human Genetics (ASHG) is the primary professional membership organization for human genetics specialists worldwide. The nearly 8,000 members of ASHG include researchers, academicians, clinicians, laboratory practice professionals, genetic counselors, nurses and others with a special interest in human genetics. The Society’s mission is to serve research scientists, health professionals and the public by providing forums to: (1) share research results through the Annual Meeting and in [The American Journal of Human Genetics \(AJHG\)](#); (2) advance genetic research by advocating for research support; (3) educate future genetics professionals, health care providers, advocates, teachers, students and the general public about all aspects of human genetics; and (4) promote genetic services and support responsible social and scientific policies. For more information about ASHG, please visit <http://www.ashg.org>.

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