



## **PRESS RELEASE**

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### **New Research Validates Clinical Use of Family Health History as the ‘Gold Standard’ for Assessing Personal Disease Risk**

***Experts Present Findings at ASHG 60<sup>th</sup> Annual Meeting that Provide New Insight on Patient and Physician Understanding and Use of Family Health History to Assess Disease Risk***

BETHESDA, MD – October 22, 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 60<sup>th</sup> 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 feature research on the application and use of family health history information in clinical settings to assess an individual's risk for developing common chronic diseases. Family health history assessment is an inexpensive, simple, and useful tool that has been shown to be effective and accurate when implemented in clinical care settings to assess personal disease risks. Integrating the use of family health history information in clinical practice can help practitioners determine which patients are at high risk of developing a specific health condition and would benefit from taking precautionary measures to prevent disease (such as early and frequent screening, genetic testing, health behavior and lifestyle changes, etc.).

To support the integration and application of family history information in clinical practice, several federal agencies and professional organizations have developed comprehensive (and free) online tools that make it easier for patients to collect, record and organize their family health history information and share it with their doctors. Some of these Web-based tools were also designed to help health care providers to correctly interpret and understand their patients’ family health history information, incorporate the data into electronic health records (EHRs), and effectively apply this knowledge in clinical practice to help determine which patients are at high risk for hereditary diseases. [*\*Note: For a comprehensive source of family health history information, online tools, and resources for consumers and health care practitioners, please see our Talk Health History Campaign Web site at: [www.talkhealthhistory.org](http://www.talkhealthhistory.org).*]

Since [National Family Health History Month](#) is celebrated in November, ASHG will be spreading awareness about this important public health topic and helping people understand its application in clinical practice as a cost-effective tool for assessing disease risk by hosting a press briefing to highlight some of the latest research findings of interest on this topic that will be presented at the ASHG 2010 Annual Meeting. The four abstracts featured in this session will present new research results that advance our understanding of how both patients and health care practitioners interpret and apply family health history information help assess personal disease risk. Two of the abstracts featured in this session will also provide new findings on the public’s use of Web-based family health history tools – including the [U.S. Surgeon General’s “My Family Health Portrait”](#) online tool and the [CDC’s Family Healthware™](#) software program – that are designed to help patients collect and record their family health history, and organize the information so that it can easily be shared with doctors and added into their EHRs.

ASHG invites members of the media to attend this press briefing titled, “New Research Validates Clinical Use of Family Health History as the ‘Gold Standard’ for Assessing Personal Disease Risk,” which will be held on Friday, November 5, 2010, from 10:30-11:30 a.m. in the ASHG Press Briefing Room at the Walter E. Washington Convention Center (Room #101). **If you wish to view the online webcast recording of this press briefing after the event, please go to: <http://www.ashg.org/familyhistoryassessment/>**

The featured speakers and research abstracts presented in this press briefing include the following:

- **Charis Eng, MD, PhD** – [“Comparison of Family Health History to Personal Genomic Screening: Which Method is More Effective for Risk Assessment of Breast, Colon, and Prostate Cancer?”](#)
- **Nedal Arar, PhD** – [“Veterans’ Response to Their Use of the U.S. Surgeon General’s Family Health History Online Tool”](#)
- **Wendy Rubinstein, MD, PhD** – [“Components of Family History Associated with Women’s Disease Perceptions for Cancer: A Report from the Family Healthcare™ Impact Trial \(FHITr\)”](#)
- **Maren Scheuner, MD, MPH** – [“Family Health History Education to Improve Genetic Risk Assessment for Cancer”](#)

Brief summaries of the major research findings reported in these four abstracts are included below:

### ***Comparison of Family Health History to Personal Genomic Screening: Which Method is More Effective for Risk Assessment of Breast, Colon, and Prostate Cancer?***

Family health history information is a useful and accurate tool for assessing an individual’s risk of developing various diseases. Family health history charts – known as pedigrees – are also an important tool that is often used in genetic evaluation. While specific personal genetic screening tests may be useful in some situations, research on family history data shows that family health history information may be a more useful initial disease risk assessment tool.

A group of researchers led by [Charis Eng, MD, PhD](#), Chair and Founding Director of the Genomic Medicine Institute (GMI) of the Cleveland Clinic, and Founding Director and Attending Clinical Cancer Geneticist of GMI’s Center for Personalized Genetic Healthcare, investigated the concordance and relevance between family health history-based risk assessment (FHRA) and SNP-based risk assessment through a DTC personal genomic screening (PGS) test. The DTC genetic tests for this study were conducted by Navigenics to assess each participant’s personal risk of developing three common types of cancer. Dr. Eng and her colleagues then compared FHRA with Navigenics PGS for breast cancer (22 females), prostate cancer (22 males), and colorectal cancer (44 individuals).

The researchers found that although both FHRA and PGS placed an average of 40% in the same risk categories, there was little concordance between the two for breast, prostate, or colon cancer risks. For instance, the use of FHRA methods classified eight individuals as being at high risk for breast cancer, but only one of the eight was classified as high-risk when assessed via PGS. Overall, FHRA assigned 22 individuals to the hereditary risk category (i.e., due to a high risk mutation in a cancer-predisposing gene), but PGS identified only one of these individuals as high-risk. The researchers also assessed nine individuals with hereditary risk for colorectal cancer, five of whom had proven mutations defining inherited colorectal cancer syndromes. None of the nine were classified as high-risk when assessed through PGS risk analysis.

“Our research findings indicate that family health history assessment is currently a better predictor of cancer risk when compared with personal genomic testing methods,” said Dr. Eng. “However, the personal genomic screening tests could potentially become more effective and accurate if combined with family health history information and used as a *complementary* tool for cancer risk assessment.”

“Evaluation of family health history still remains to be the gold standard in personal disease risk assessment,” said Eng. “Until further research is conducted to prove that personal genomic screening can accurately predict a person’s risk, it is imperative that health care practitioners rely on family history as the primary tool that they use to clinically evaluate each patient’s personal risk of developing cancer.”

Furthermore, Eng points out that, "A personal family health history report can be completed at little to no cost, and this type of information is typically readily accessible and easily gathered by the patient." She concluded by stating her belief that, "In effect, family health history could potentially be considered as a 'secret weapon' in the integration of personalized medicine and genetic knowledge into clinical practice."

### ***Veterans' Response to Their Use of the U.S. Surgeon General's Family Health History Online Tool***

Despite growing evidence regarding the importance and efficacy of using detailed family health history as a key tool in common disease risk assessment, this type of approach for screening and identifying at-risk individuals has not been broadly applied in clinical practice yet. To address this problem, a group of researchers led by [Nedal H. Arar, PhD](#), Associate Professor of Medicine at the University of Texas Health Science Center at San Antonio and a research scientist at the South Texas Veterans Health Care System, built on their collective experience in genetic family studies and genomic health service research to study processes and ethical issues associated with use of the [U.S. Surgeon General's "My Family Health Portrait" online family health history tool](#) by veterans. In the current study, Dr. Arar and her colleagues assessed veterans' satisfaction in using the U.S. Surgeon General's online family health history (SG-FHH) tool, and their intentions regarding the utilization and application of their family health history (FHH) information in clinical settings.

A total of 35 veterans who were patients at the primary care outpatient clinic in San Antonio, Texas, were asked to enter their FHH information into the online SG-FHH tool, and complete the study's surveys. The study participants were given a printed copy of their family history pedigree that was based on the data that they entered into the online SG-FHH tool. They were then asked to participate in a brief, semi-structured interview to assess their intentions of how they are planning on using their FHH information. The participants were also asked to fill out a survey that assessed drivers of satisfaction (i.e., content, functionality, look and feel, navigation, and performance) in using the SG-FHH online tool.

The results of this study indicate that all 35 participants found the online tool to be very useful. About 67% of the participants said that they have easy access to a computer or the Internet and demonstrated their ability to complete all FHH forms. Most of the participants (88%) viewed the functionality, look and feel, navigation, and performance of the SG-FHH tool favorably. However, content analysis of the semi-structured interviews indicated that there are several barriers to veterans' use of FHH information, including their lack of knowledge regarding the importance of familial risks in developing chronic diseases, and privacy and confidentiality concerns related to entering their personal data into an online database.

Overall, this study provided a positive assessment of the performance and functionality of an inexpensive and widely-accessible method for collection and documentation of FHH information that can be integrated into clinical practice to help practitioners determine which patients are in need of more rigorous early disease screening, genetic testing, and/or special preventive measures and therapies. The study findings also highlight several opportunities and challenges related to the utilization of FHH information as a clinical and genomic tool.

"Our work makes a substantial contribution to the ongoing efforts directed toward the integration and translation of genomic research findings into clinical practice," said Dr. Arar. "The results of our study suggest that promoting the use of Web-based family health history screening tools such as the Surgeon General's could have important implications for primary care providers and their patients. Increasing the public's use of this type of online tool would improve screening for common complex diseases, which would allow health care providers to focus their resources on addressing critically important health behaviors and preventative measures in the populations that are at highest risk for chronic diseases."

She also noted that, "Overall, successful implementation of family health history information in clinical practice has significant implications for personalized medicine. It will also reduce health care costs by improving disease risk screening and prevention, which will, in turn, help improve health and quality of life for many Americans."

## **Components of Family History Associated with Women's Disease Risk Perceptions for Cancer: A Report from the Family Healthware™ Impact Trial (FHITr)**

The [Centers for Disease Control and Prevention \(CDC\)](#) has developed a Web-based predictive disease risk software program called [Family Healthware™](#) as a tool to help evaluate an individual's familial risk for heart disease, stroke, diabetes, and colon, breast, and ovarian cancer.

[Wendy S. Rubinstein, MD, PhD](#), Clinical Associate Professor of Medicine at the University of Chicago and Director of the NorthShore University HealthSystem Center for Medical Genetics, and her colleagues at the University of Michigan and Case Western Reserve University received a three year grant from the CDC to evaluate this new Web-based family history tool and report on the impact of the program's familial risk report and health messages on users' health behaviors, attitudes, and their use of health care services. The current study was designed to determine those elements of family health history and personal characteristics most closely related to women's risk perceptions about cancer.

Dr. Rubinstein's team examined data from 2,505 healthy women, ages 35-65, who were enrolled through 41 primary care practices participating in the [Family Healthware™ Impact Trial \(FHITr\)](#). The women reported detailed information about their family health history of coronary heart disease, stroke, diabetes, and breast, colon, and ovarian cancer. In addition, the women also reported on their perceived risk, extent of worry, perceived severity and perceived control over getting (or preventing) these diseases.

The study participants provided information on a total of 41,841 relatives. Excluding their parents, significantly more women reported having a family history of cancer in their maternal versus paternal lineage. In addition, having a family history of cancer on the paternal versus the maternal side was associated with a lower perceived risk for breast cancer. Cancer-specific perceived risks were present for all three cancers for the total number of relatives, or having any first-degree relative with that type of cancer. The research results also indicate that there were novel "spillover" effects wherein a positive family history of breast or colon cancer was associated with a higher perceived risk of ovarian cancer. Furthermore, according to the study findings, age and parenthood were associated with disease perceptions in ways that ran counter to empiric risks.

The researchers concluded that gaining a better understanding the elements that contribute to women's disease perceptions has important implications for communicating health risks to this target audience. The research findings suggest that female patients have limited awareness and understanding of the relevance of their paternal family medical history. Some have suggested that public health education programs should place a stronger emphasis on patient-centered approaches aimed at helping patients achieve informed choices instead of generally promoting the positive value of early disease screening, genetic testing, or preventive surgeries and therapies. The current research findings support this approach and identify key areas of women's disease risk perceptions that should be considered when developing public health messages for this target audience.

"The experience of living in a family shapes our attitudes toward illness and health, which in turn exerts a large influence on whether or not we adopt healthy behaviors – not to mention the fact that our relatives tend to share a lot of genetic overlap in terms of the disease risks that we have inherited," said Rubinstein. "It is for these reasons that family health history is considered to be the 'gold standard' for disease risk assessment that will help us achieve the full benefit of personalized medicine through the effective use and application of patients' genetic information in clinical practice."

"The results of our analyses indicate that while health risk perceptions are generally in agreement with medically-recognized family history risk factors, some perceptions often run counter to actual risk," Rubinstein noted. "For example, our research findings suggest that many patients think cancer risk decreases with age, when in fact it *increases* with age."

"Understanding women's 'mental model' of disease and how they interpret family health history information is critical for effective communication of disease risk through health care providers, family history tools, and public health campaign messages designed for this target audience," said Rubenstein.

## ***Family Health History Education to Improve Genetic Risk Assessment for Cancer***

Implementation of a comprehensive genetics education program for primary care clinicians at the Department of Veterans Affairs Greater Los Angeles (VA GLA) Healthcare System has resulted in better cancer family history documentation, improved recognition of patients at risk for hereditary cancer, and increased referrals for genetic consultation and testing.

This education program – which was developed by [Maren Scheuner, MD, MPH](#), Director of the Health Services Genomics Program and Clinical Genetic Services at VA GLA, and a group of her colleagues – is comprised of informational (Web site, CME-approved lecture series), clinical (patient and provider information sheets, a family history reminder with template in the electronic health record (or EHR)), and behavioral components (practice-feedback reports regarding clinicians' use of the EHR tool).

To evaluate the impact of the education program, Scheuner and colleagues kept track of changes in the knowledge, attitudes and behaviors of seven clinicians (five physicians, one nurse practitioner, and one physician assistant) who practice in the Women's Clinics at VA GLA and have agreed to participate in the study. At baseline, the mean percent correct score for clinician knowledge was 55% (range: 26-77%) with the lowest scores in the areas of genetic testing and recognition of hereditary cancer syndromes. The topics rated as most relevant to primary care practitioners were management of hereditary cancer, knowing when to refer a patient, and ethical issues.

The clinical tools developed for the EHR appear to have had the greatest impact in changing clinician behavior. During the first five months of implementation of the education program, 568 cancer family histories were completed using the reminder and template in the EHR. The family history data captured with the template includes information that previously was not routinely documented by the clinicians, but is necessary to recognize high-risk patients, such as a family history of cancer in second-degree relatives, identifying the maternal or paternal lineage of affected relatives, and the age of cancer diagnosis in relatives. As a result, the genetics consultation service at the VA GLA received 47 patient referrals for risk assessment and possible genetic testing that, according to Scheuner, "very likely would not have been made without this comprehensive genetics education program."

"Our early experience with this education program shows that we have successfully changed the behaviors of our primary care clinicians at the VA in Los Angeles," Scheuner said. "Our research results suggest that the educational components implemented in this program have helped improve family health history documentation and implementation on the part of clinicians, which likely played in a large role in contributing to the subsequential increase that we saw in the number of patients who were referred to the VA GLA genetics consultation service for risk assessment and/or genetic testing."

However, she noted that evaluation of the genetics education program is still ongoing. "We are planning to conduct interviews with the clinicians enrolled in this study to get feedback about the individual components of the program, and obtain a follow-up assessment of the clinicians' knowledge and attitudes about genetics, family history documentation, and risk assessment," said Scheuner. "If we demonstrate continued success, this program may serve as a model for improving genetic risk assessment of other health conditions, and with appropriate modifications, it could also potentially be applied in other health care settings as well."

## ***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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