



PRESS RELEASE

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GENETIC ANCESTRY TESTING CHALLENGES IDENTIFIED BY AMERICAN SOCIETY OF HUMAN GENETICS TASK FORCE

ASHG Publishes New White Paper in Journal that Explains Scientific Reasoning and Expands on Recommendations Made in 2008 Policy Statement

BETHESDA, MD – May 13, 2010 – Genetic ancestry testing is a practice that has become increasingly popular in the U.S. over the past few years. An estimated half-million Americans will purchase genetic ancestry tests from one of the approximately 40 companies worldwide that currently provide this type of service to consumers. However, increasing public interest in Direct-to-Consumer (DTC) genetic ancestry testing has been accompanied by growing concern about the personal and societal implications of this type of testing, and issues related to the scientific validity of ancestry inference methods.

Ancestry estimation has enormous value in human genetics research when used to reveal patterns of past human migration and to provide a background for emerging patterns of human genetic variation. However, ancestry is often imprecisely defined and identified, and researchers are lacking a standard set of guidelines for best practices.

There has been a recent proliferation of companies that provide genetic ancestry testing and an increasing number of individuals who are interested in using these tests to confirm or extend their knowledge of family genealogy. However, genetic ancestry tests remain unregulated with no oversight, nor industry guidelines to ensure the quality, validity, and interpretation of these tests. Concerns regarding the accuracy of genetic ancestry inference and the opportunity for harm associated with misinterpretation of genetic ancestry data must be addressed.

Scholars and scientists have urged the genetics community to address these issues and take on a leadership role in offering guidance for DTC genetic ancestry testing and to provide standards for testing. The [American Society of Human Genetics \(ASHG\)](#) considers issues related to the scientific accuracy and direct-to-consumer marketing of genetic tests to be a priority area, as demonstrated by their statements on [DTC genetic testing \(health-related\)](#) and [genetic ancestry testing](#).

In an effort to address issues specifically related to genetic ancestry testing, ASHG established an Ancestry Testing Task Force Committee that was composed of some of the world's top scholars in this research area. In November 2008, the Task Force Committee provided the [ASHG Ancestry Testing Summary Statement and Recommendations](#) at the Society's 58th Annual Meeting in Philadelphia. [NOTE: The 2008 [ASHG Ancestry Testing Statement](#) and [press release](#) are available on the Society's Web site.]

In responding to the stated needs put forth in the 2008 Ancestry Testing Statement, the ASHG Task Force has published a comprehensive White Paper in the May 14, 2010 issue of [The American Journal of Human Genetics \(AJHG\)](#). This report – titled, [“Inferring Genetic Ancestry: Opportunities, Challenges, and Implications”](#) – provides a more in-depth analysis of key scientific and non-scientific aspects of genetic ancestry inference that are prevalent in academia and industry. The report also offers recommendations for advancing the current debate

and facilitating the development of scientifically based, ethically sound, and socially attentive guidelines concerning the use of these continually evolving technologies.

"This report not only expands upon the Task Force's 2008 summary of the benefits, limitations, and potential outcomes of genetic ancestry estimation in research and industry, but it also suggests specific action items for helping to accomplish the ideals put forward in the ASHG ancestry testing statement," said Charmaine Royal, Ph.D., lead author on the *AJHG* White Paper and co-chair of the ASHG Ancestry Testing Task Force Committee.

In their 2008 statement, the Task Force stated that the ASHG views academia, industry, and consumers as sharing responsibility for conveying and understanding the limitations of genetic ancestry testing. The ASHG Task Force also stressed the importance of engaging the scientific community and industry, to ensure that these parties fully inform consumers about the limitations, risks, and benefits involved in genetic ancestry testing.

Based on their review of the current state of the science and the personal, societal, and health-related implications of genetic ancestry inference in academia and industry, the ASHG Ancestry Testing Task Force has made the following recommendations in their report published in *AJHG*:

- 1) Leaders of the human genetics community should develop mechanisms for promoting thoughtful and rigorous use of genetic ancestry estimation in academic research.**
- 2) Interested scientific and scholarly societies should collaborate to convene a national roundtable discussion of DTC genetic ancestry testing.**

According to the ASHG Task Force's White Paper report, effective decision-making regarding genetic ancestry inference – and DTC genetic ancestry testing in particular – will be best initiated through cooperative interaction among a variety of stakeholders, including scientific societies, industry, and suitable federal agencies.

The goal of coordinating a face-to-face discussion among these groups would be to identify major issues of concern and brainstorm practical solutions. Some of the key points that must be considered by participants in this discussion include: scientific accuracy and the reporting of statistical confidence, proprietary databases, additional research that must be conducted, interdisciplinary collaboration, suggestions and mechanisms for industry accountability, communication of test limitations and potential consequences, and public and personal education about these issues.

"ASHG has agreed to take on a leadership role in response to the recommendations made in the Task Force's White Paper report," said [Joann Boughman, Ph.D., Executive Vice President of ASHG](#). "The recommendations included in this White Paper are intended to promote an open and collaborative discussion about the key issues in genetic ancestry inference that are prevalent in academia and industry."

"The issues and solutions discussed at a face-to-face meeting of the key stakeholders can inform important decisions about next steps," said Boughman. "It is our hope that engaging these groups in an inclusive and productive dialogue will move us closer to identifying and addressing the major issues of concern in genetic ancestry inference."

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 involved in or 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/>.