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ASHG 2008: Recommendations Issued on Ancestry Testing

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November 14, 2008 (Philadelphia, Pennsylvania) — The American Society of Human Genetics (ASHG) released an Ancestry Testing Statement here yesterday during their 58th Annual Meeting. The ASHG statement considers commercial ancestry testing, ancestry estimation, motivations for assessing ancestry, accuracy of the tests, and the health, personal, and societal implications of the tests.

An earlier ASHG statement dealt with direct-to-consumer genetic testing specifically related to healthcare or decisions. The new statement was motivated by the proliferation of companies that offer both health and ancestry genetic information, and by the psychological, social, political, legal, ethical, and health-related effects of the test results. Genetic tests of ancestry provide valuable information about human migration and human genetic variation, with results based on statistical probabilities. However, individuals interested in genetic ancestry testing often want information more precise than these tests can provide.

Several members of the ASHG Ancestry Task Force Committee were introduced at a press briefing by the current president of ASHG, Aravinda Chakravarti, PhD, professor at Johns Hopkins University School of Medicine Institute of Genetic Medicine in Baltimore, Maryland, and ASHG President-Elect Edward McCabe, MD, PhD, professor and executive chair, Department of Pediatrics, David Geffen School of Medicine, and physician-in-chief, Mattel Children's Hospital, University of California in Los Angeles.

Committee's Hard Work

"I was an observer," said Dr. McCabe of his role. "I'm not a population geneticist or a mathematical geneticist, but this is a committee that really got down and did a lot of hard work, and this is really the beginning of the work of the committee that we're going to be discussing.... They will be putting together a white paper, a much longer, more scholarly document, which will be coming out in the spring."

The new ASHG statement includes 5 recommendations:

- Industry and academia should clarify the limitations of ancestry testing for consumers, and consumers should use resources available to understand the meaning and limitations of ancestry testing
- Research is needed to understand how sampling, geographic patterns of diversity, marker selection, and statistical analysis affect test accuracy
- Consequences of test results should be assessed, and explanation or counseling should be available in direct-to-consumer, healthcare, and research settings
- Collaboration is needed between scientists involved in genetic ancestry studies and scholars versed in cultural, historical, social, and political areas
- Increased accountability should be explored for companies offering direct-to-consumer ancestry testing

Committee member Michael Bamshad, MD, professor of pediatrics and adjunct professor of genome sciences at the School of Medicine, University of Washington, Seattle, explained, "When we talk about ancestry testing, we're talking about using a set of genetic markers to make inferences about an individual's ancestry. And there are many types of marker systems that can be used."

Potential Marker Systems

The most familiar are mitochondrial DNA markers, transmitted from a woman to her offspring, and markers on the Y-chromosome

transmitted from fathers to sons. More recently, people are using markers distributed across the genome — single nucleotide polymorphisms (SNPs). "Any of these," said Dr. Bamshad, "can be used as ancestry-informative markers." A marker that has quite different allele frequencies in different populations may provide information about ancestry. In ancestry testing, companies use batteries of these markers to draw inferences about origins.

"[People] have the expectation that [ancestry testing] is going to provide them with presumably very accurate information about their recent ancestry," Dr. Bamshad said. Ancestry testing depends on making inferences about human variation, but "even in the best databases that exist today, we know that we have only a very limited sampling.... There are large gaps in our knowledge about human genetic variation," he noted.

Committee co-chair Charmaine Royal, PhD, from the Institute for Genome Sciences & Policy, Duke University, Durham, North Carolina, described some implications of ancestry testing. "Geneticists study genes...and that's the part of the ancestry that they're primarily interested in," she said. "But there's a lot of concern that the focus on genetic ancestry will minimize the focus, or the importance, of the social factors, the cultural factors, and the political and legal factors that also impact who we are and how we define ourselves."

Dr. Royal continued, "Scientists who are doing research in population genetics, looking at how populations are related, people in the commercial enterprises doing ancestry testing — it would behoove them to connect with and talk with people in the social sciences."

Political and Legal Implications

Another concern is that people are using the results of these tests to claim benefits associated with membership in a certain group such as Native Americans or African Americans. "There are questions about the political and the legal implications of this information, and what is appropriate use of this information," Dr. Royal said.

Medscape Pathology & Lab Medicine asked whether ancestry testing is able to determine the specific ethnic group from which a person is descended.

"Genetic testing can give them some information about their maternal or paternal lineage as they relate to population groups," said Dr. Royal. "One thing about the populations, speaking of Africa in particular...the groups in the databases are not the pre-Middle Passage ancestors necessarily. Those names have probably changed over time, and people have moved to different parts. So the information that they're getting now, and the group names that they're getting, might be different from what the actual groups were."

"None of the ancestral populations for which testing is being done are explicitly in those databases," Dr. Bamshad agreed. "For example, when we talk about West African ancestry, we're using a contemporary West African population to represent the ancestors of contemporary African Americans."

Potential Usefulness of Tests

Medscape Pathology & Lab Medicine spoke with Wayne W. Grody, MD, PhD, professor in the Divisions of Medical Genetics and Molecular Pathology, Departments of Pathology and Laboratory Medicine, Pediatrics, and Human Genetics, School of Medicine, University of California, Los Angeles, after the press briefing.

"We've all become concerned lately about the great expansion of direct-to-consumer marketing and ordering of genetic tests, and these even include genetic disease tests," said Dr. Grody. "Of course, this press conference was about strictly ancestry, but many of the markers are the same — the same markers we might associate with descent from a certain region could also go along with certain genetic diseases that came from that same region. And that's why several of the speakers referred to the potential importance or usefulness of this in healthcare."

"I think the take-home message is, the distances and time that we're talking about are so huge," Dr. Grody noted. "And we have no way to sample the actual origins we're trying to conclude. By the time it gets down to us, it's quite nebulous, and all you get are some probabilities. So personally I wouldn't put a huge amount of stock into it, other than maybe satisfying a bit of curiosity."

Dr. Bamshad, Dr. Royal, and Dr. Grody have disclosed no relevant financial relationships.

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