



ASHG Urges Closer Look at Consumer Ancestral DNA Tests

November 13, 2008

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PHILADELPHIA (GenomeWeb News) – Taking a DNA test to learn about one’s ancestry is becoming more popular, and this branch of consumer genomics is beginning to blend with other direct-to-consumer genotyping services, leading to concerns that the implications of such testing have not been adequately addressed, according to the American Society of Human Genetics.

In response to this concern, ASHG today issued a set of [recommendations](#) to the roughly 30 players in the ancestral genetics testing field and to academics at its annual meeting here.

The society said it believes that ancestry testing “warrants independent consideration” because an increasing number of companies are offering both ancestral and health-related tests, and because the impact of these tests on families and communities “traverses a wide range” of ethical, cultural, health-related, and other issues. ASHG also said that many of the scientific and non-scientific challenges and implications of DTC ancestry testing are not being adequately addressed in the research communities that gave rise to these technologies.

“The applications and uses of ancestry assessment are quite different when implemented for the purposes of scientific research on population genetics, versus the commercial applications of this type of test as a service that individual consumers can purchase to learn more information about the ethnic and geographical origins of their ancestors,” ASHG President Aravinda Chakravarti said in a statement today.

“The distinction between the different applications of ancestry assessment can be a source of confusion and misunderstanding for both consumers and researchers,” Chakravarti added.

Because of the limitations of the science of determining ancestry, greater efforts are needed to make these limits clearer to consumers, ASHG advised in a paper it presented to the annual meeting. The public also bears the responsibility of studying the information about these tests and to “strive to better understand the implications and limitations of these assessments,” it said.

There are numerous potential implications of ancestry testing, Charmaine Royal, of the Duke University Institute of Genomic Sciences and Policy, said during ASHG’s press conference today.

For instance, she noted, ancestry overall involves a complex interplay between genes, society, and culture. That raises concerns that genetic studies will minimize the importance of these social, cultural, and political factors. For her part, Royal said that she feels very strongly that commercial ancestry testing companies should communicate with other groups to take such factors into account.

And, Royal said, there are also concerns that some individuals will use ancestry information to try to make claims to certain benefits. “There are questions about the political and legal implications of this information,” she said.

In addition, it is possible that test results could have a psychological impact on people who question their identity based on the results, Royal said. “People respond to the information — they use it — differently,” she said, adding, “The test does not have to be inaccurate to cause problems.”

Mechanisms for greater accountability of direct-to-consumer genetic testing should be explored, according to the ASHG recommendations. The group did not offer details about what sort of mechanisms, but said that in a commercial environment, accountability could be “further compromised by various market pressures.”

The organization also said that more studies are needed to better understand how the accuracy of genetic ancestral information is influenced by who has been sampled in the databases, by human geographical patterns, and by marker selections and statistical methods.

Ancestry-informative markers have been used over the past two decades or more to make inferences about human evolution and demography, according to Michael Bamshad of the University of Washington, who also spoke at ASHG's press conference. They can also be used as a tool in an attempt to refine interpretations of genetic susceptibility tests in different populations, he added.

In contrast to using ancestry testing as a tool, Bamshad added, it can also be a product itself. Consumers “have an expectation that it's going to provide them with, presumably, very accurate information about their recent ancestors,” Bamshad said.

Even so, he added, the accuracy of tests depends on a number of factors, including the data available about reference populations, inference approaches, and so on. And, Bamshad said, each marker provides a different level of robustness and power, meaning which marker system you use — and how many markers — makes a difference as does the statistical analysis used to interpret the information.

ASHG also thinks that there ought to be guidelines about how to counsel people on how ancestry estimation works in terms of direct-to-consumer programs, in research, and in healthcare settings. In addition, it said that scientists inferring genetic ancestry should consult or collaborate with scholars with knowledge about the historical, sociopolitical, and cultural contexts that could “inform the processes and outcomes of their research and commercial efforts.”

“Consumers, as well as scientists, must remember that ancestry-testing inferences are fallible, and that over-interpretation or misinterpretation can happen,” explained Edward McCabe, ASHG's president-elect. “Inaccurate results may be confusing and life-changing, therefore greater efforts are needed to make the limitations of ancestry testing more explicit.”

Implementing these recommendations “is likely to have many benefits,” ASHG said. The payoffs include improved knowledge of human evolution and demographic history, more accurate testing, “better informed users of ancestry information, and the establishment of a framework for interpreting ancestry information in a culturally appropriate and socially sensitive manner.”

Additional reporting for this article was done by Matt Jones in New York.