

## American Society Of Human Genetics Releases Statement On Genetic Ancestry Testing At 58th Annual Meeting

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This year, an estimated half-million Americans will purchase genetic ancestry tests at costs ranging from \$100 to nearly \$1,000 per test. With the growing popularity of direct-to-consumer (DTC) genetic testing, it is imperative to explore the complex relationship between genetics, ancestry and race that can complicate the interpretation of these types of tests and their results.

Ancestry estimation can have enormous value in human genetics research, revealing patterns of past human migration and providing a background pattern of human genetic variation, but it is often imprecisely defined and identified. In addition, any individuals are interested in using genetic ancestry testing to confirm or extend their knowledge of family genealogy, but current limitations regarding the accuracy, appropriate interpretation, and opportunity for harm associated with misinterpretation of the data, must be acknowledged.

Those who undergo ancestry testing often do not realize that the tests are probabilistic and can reach incorrect conclusions, causing emotional distress if test results are unexpected or undesired. Consumers frequently purchase these tests to learn about their race or ethnicity, yet there is no clear-cut connection between an individual's DNA and racial affiliation. These tests are also being referenced in medical settings as the public becomes more aware of the association between ancestry and disease. Patients may ask doctors to take their ancestry tests into consideration when making medical decisions, although the results are often inconclusive.

A recent ASHG statement on DTC genetic testing acknowledged the prominence of commercial ancestry testing, but focused explicitly on tests that make health-related claims or that directly affect health care decision making. However, the Society believes that ancestry testing warrants independent consideration for the following reasons: 1) an increasing number of DTC genetic testing companies are offering both ancestry and health-related genetic information; 2) the impact of ancestry testing on individuals, families and communities traverses a wide range of ethical, legal, social and health-related issues; and 3) many scientific and non-scientific challenges and implications of ancestry testing affect both commercial applications and genetic research efforts in this area.

Members of the ASHG Ancestry Testing Task Force Committee, including current president [Aravinda Chakravarti, Ph.D.](#), and president-elect, [Edward McCabe, M.D., Ph.D.](#), will unveil the Society's ancestry testing recommendations in a press briefing session at the [ASHG 58th Annual Meeting](#) in Philadelphia on Thursday, November 13, 2008. (See press briefing session and webcast details below.)

"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," said ASHG President Aravinda Chakravarti, Ph.D., an expert in population genetics at Johns Hopkins University School of Medicine.

"The distinction between the different applications of ancestry assessment can be a source of confusion and misunderstanding for both consumers and researchers," Chakravarti explained. "However, ASHG believes it is extremely important that the scientific research community, industry and the public understand the limitations, risks and benefits involved."

For consumers, personal decisions about ancestry testing may ultimately depend on the cost and type of genetic markers used in the test, as well as the specificity of the information that the test results are designed to provide. The ASHG statement cautions consumers that they need to be aware of and informed about the practical limitations in the accuracy of ancestry test results. According to the statement, the accuracy of this type of testing greatly depends on the context and statistical methods used to perform ancestry estimation, regardless of the type of marker systems utilized.\*\*

Furthermore, the terms "race" and "ethnicity" are typically used to categorize the human population into separate subgroups may be intertwined with ancestral origins, and these delineations are

sometimes considered to be a key determinant of disease risk. However, it is unclear whether race may be associated with disease susceptibility due to biological or genetic factors, or whether such associations exist because these features result from environmental factors. Many researchers believe that different combinations of both genetic and environmental factors play a role in influencing human disease risk. Thus, although there are circumstances in which the genetic factors influencing health-related traits do tend to vary among racial groups, it is unclear whether (and to what extent) such risk factors account for variation in the prevalence or direct causation of these traits among different subgroups of the population.

While some challenges in research-related ancestry testing overlap with individual and commercial ancestry testing, certain issues become highlighted in the consumer testing world. For some groups (certain Native American tribes, for example), a major concern about scientific and commercial efforts to explain ancestral origins is the apparent lack of regard for important cultural, religious, social, historical and political processes that also inform group origin, membership, identity, and access to group rights. The reshaping of individual or group identity can elicit a range of psychological responses, and may result in confusion or distress, especially in the situation of conflicting information.

### **ASHG Ancestry Testing Recommendations**

The Society's ancestry testing recommendations for the scientific community, industry, consumers, and the general public include the following:

1. Greater efforts are needed on the part of both industry and academia to make the limitations of ancestry testing estimation more clear to consumers, the scientific community, and the public at large. In turn, the public (consumers, in particular) have a responsibility to avail themselves of information about ancestry testing and strive to better understand the implications and limitations of these tests.
2. Additional research is required to further understand the extent to which the accuracy of genetic ancestry estimation is influenced by the individuals represented in existing databases, geographical patterns of human diversity, marker selection and statistical methods.
3. The complex consequences of ancestry testing for individuals, families, and populations need to be assessed and guidelines should be developed to facilitate explanation and counseling about this type of testing in scientific research, DTC and health care related settings.
4. Scientists inferring genetic ancestry should consult or collaborate with scholars who have expertise in the historical, sociopolitical and cultural contexts needed to inform the processes and outcomes of their research and commercial efforts.
5. Mechanisms for greater accountability of the DTC ancestry testing industry should be explored.

"In writing this statement, a guiding principle of our work has been that people have an intrinsic interest in their ancestry, and the human genetics community encourages this interest, as we celebrate the diversity among us," said Charmaine Royal, Ph.D., co-chair of the ASHG Task Force Committee. "However, the desire to learn about ancestry should be coupled with a drive to understand the subtleties of the problem."

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

### **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>

\*\*The types of markers used in genetic ancestry testing may be patrilineal, as is the case with tests that use Y chromosome markers; matrilineal, as is the case with tests using mitochondrial DNA markers; and some of the more recent genetic tests use markers that span a number of different areas across the entire human genome, as is the case with tests that use single nucleotide polymorphisms (SNP) markers. A subset of SNP markers that are commonly referred to as Aims (ancestry informative markers) may also be used.

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