Personal Genetic Ancestry Testing: Concerns Regarding Accuracy

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The ability to trace one's ancestry with a simple test is an alluring proposal for many. This can be particularly interesting for people who have been impacted by diasporas and lack a clear picture of their bloodlines. According to Dr. Troy Duster[1], genetic ancestry tests are conducted in multiple ways: using people's mitochondrial DNA (mtDNA) testing, Y-chromosomes, and autosomal markers. These methods can be employed — either by themselves or in conjunction — to offer an approximation to where a person's ancestors lived (Duster, 2011). These methods are used by many companies which sell genetic ancestry testing. However, concerns over its accuracy have been called into question by countless researchers. Despite the concerns, companies, such as 23andMe and African Ancestry, which sell genetic ancestry tests, minimize or ignore the potential inaccuracies claiming, "to tell consumers where their ancestral lineage originated and the social group to which their ancestors belonged" (Bolnick et al., 2007). Even though personalized genetic ancestry companies claim that their products are highly accurate, this statement is highly problematic as "(i) the tests can have a profound impact on individuals and communities" (Bolnick et al., 2007)[2]. The question of their accuracy is critical. Inaccurate testing can negatively affect people's perception of themselves and how they fit in their communities. Potential limitations of personalized genetic ancestry testing are also not common knowledge, so tests can significantly impact people's lives without them realizing the potential for mistakes.

Personalized genetic ancestry testing is done through 3 methods; two of these methods, mtDNA testing and Y-chromosome testing, function similarly. Both look at people's haplogroups, alleles that are inherited from only one parent. The mitochondria, the powerhouse

[1] Troy Duster is the past president of the American Sociological Association
[2] Deborah Bolnick is part of the Department of Anthropology at the University of Texas, Austin

of the cell, have their own separate DNA called mitochondrial DNA (mtDNA) that is only inherited from the mother. This defines one's maternal lineage. Conversely, the Y-chromosome, the male sex chromosome, is only passed down by the father and defines one's paternal lineage (Shriver et al., 2004)[3]. These two forms of DNA can be analyzed to determine how closely two people are related to one another and to look for potential places of origins. Analyzing people's mtDNA has been leveraged by private laboratories to infer people's geographical region by matching their mtDNA to the mtDNA found in people in specific regions to try and find similarities (Emery et al., 2015). This requires the challenging task of accumulating data on the mtDNA of people in every region a company wishes as a possible result of their tests. To give comprehensive global results, data in many isolated regions will need to be found. Already, this creates a potential for inaccuracy as the more specific a company wishes to report, the more data they need. Additionally, as explained by Leslie Emery[4], mtDNA ancestry testing has a high margin of error because:

> [c]ontinental-ancestry proportions often varied widely among individuals sharing the same mtDNA haplogroup… Prediction of an individual's mtDNA haplogroup from his or her continental-ancestry proportions was often incorrect. Collectively, these results indicate that for most individuals in the worldwide populations sampled, mtDNA-haplogroup membership provides limited information about either continental ancestry or continental region of origin. (Emery et al., 2015)

mtDNA is not always unique among different areas, causing some individuals to be matched to the wrong geographical region. When it is used, mtDNA does not provide as comprehensive of a result as companies claim. The other method, Y-chromosome ancestry testing, has one obvious

[3] Mark D. Shriver is part of the Department of Anthropology at Penn State University
[4] Ph.D. in Genome Sciences at the University of Washington and part of the Department of Genome Sciences

issue: it is only found in males. Therefore, if a female wants to take the test, they require a male on their father's side to take the test for them (*African Ancestry*, n.d.). While this alone does not cause any inaccuracies, it is still a large barrier making them inconvenient for people to get. Y-chromosome tests, like mtDNA, can only trace one lineage: the paternal. It will "identify the father, but also the father's father, and if the data were available, the father's father's father" (Duster, 2011). Like mtDNA testing, this does not analyze the target's entire DNA and will not report one's full ancestry. Consequently, both tests are limited and will only be able to analyse a tiny fraction of the test-takers' DNA and only find one ancestor per generation (Bolnick et al., 2007). Even assuming perfect accuracy of these tests, they are only able to find one ancestor every generation and will not give a comprehensive view of an individual's ancestry. In all, these test results may be inaccurate and present only a small fraction of one's full ancestry.

The third method is through autosomal markers. This strategy "attempts to provide a better measure of overall ancestry by using 175 autosomal markers (inherited from both parents) to estimate an individual's 'biogeographical ancestry'" (Bolnick et al., 2007). These markers are found on various chromosomes all over people's DNA, not just the Y-chromosome and mitocondria, leading to a more comprehensive result. They provide more information about an individual's ancestry because they are found on a much larger part of their genome (Royal et al., 2010). Autosomal marker tests look at far more than both mtDNA and Y-chromosome tests, however, they are still flawed. In short, companies require large amounts of data on the genomes of people from all the regions around the world. Even tens of thousands of samples in one region might fail to account for all the genetic diversity in a population (Bolnick et al., 2007). The samples collected by companies also need to be those that are similar to the historical makeup of the region in order to provide accurate data on one's history. This presents a problem as,

"present-day patterns of residence are rarely identical to what existed in the past" (Bolnick et al., 2007). In addition, the variation in one's genome that is kept from one ancestor usually halves every generation, limiting the amount of information retained (Royal et al., 2010)[5]. This makes it harder and harder to report on one's ancestry every generation further one goes. One completely different flaw that only affects people whose parents are from different races is that autosomal marker tests will claim that they are from one single place of origin, "e.g., an individual with an East Asian and European parent will be indistinguishable from an individual from Central Asia" (Royal et al., 2010). This is an undeniable flaw in this approach of ancestry testing and will affect an increasing number of people as societies become more fluid. Despite the benefits over mtDNA and Y-chromosome testing, looking at autosomal markers is not accurate enough to give an accurate, comprehensive picture of one's ancestry.

The evidence above reveals that genetic ancestry tests are not accurate; however, this goes against the claims of numerous companies in the field. In all, these companies "offered 151 products or packages, consisting of 41 mitochondrial DNA (mtDNA) products, 57 Y-chromosome products, 28 autosomal products, 22 packages that combine mtDNA and Y-chromosome products, and three packages that combine mtDNA, Y-chromosome, and autosomal products" (Wagner et al., 2012)[6]. Given the many companies, many with their own private databases, tests from different companies can disagree causing discrepancies between their respective results. Dr. Duster presented an example of someone who took multiple tests from three different companies: African Ancestry, Relative Genetics, and Trace Genetics. On their first test, they were told their ancestors were Mende from Sierra Leone. The second test gave a different result: the Wobe tribe in the Ivory Coast. A third test stated they were from

[5] Charmaine D. Royal is part of the Institute for Genome Sciences & Policy at Duke University
[6] Dr. Wagner is part of the Center for the Integration of Genetic Healthcare Technologies at the University of Pennsylvania

Senegal (Duster, 2011). Inconsistencies between these ancestry test results disprove the companies' claims of validity and accuracy. While genetic ancestry is flawed, people still use it. As stopping people from taking these tests is an impossible task, the best solution to this issue is to educate people on the potential inaccuracies and research for accurate testing methods.

References

Bolnick, D. A., Fullwiley, D., Duster, T., Cooper, R. S., Fujimura, J. H., Kahn, J., ... & TallBear, K. (2007). The science and business of genetic ancestry testing. *Science*, *318*(5849), 399-400.

Royal, C. D., Novembre, J., Fullerton, S. M., Goldstein, D. B., Long, J. C., Bamshad, M. J., & Clark, A. G. (2010). Inferring genetic ancestry: opportunities, challenges, and implications. *The American Journal of Human Genetics*, *86*(5), 661-673.

Duster, T. (2011). 5. Ancestry Testing and DNA. In *Race and the Genetic Revolution* (pp. 99-115). Columbia University Press.

Emery, L. S., Magnaye, K. M., Bigham, A. W., Akey, J. M., & Bamshad, M. J. (2015). Estimates of continental ancestry vary widely among individuals with the same mtDNA haplogroup. *The American Journal of Human Genetics*, *96*(2), 183-193.

Jorde, L. B., & Bamshad, M. J. (2020). Genetic ancestry testing: What is it and why is it important?. *Jama*, *323*(11), 1089-1090.

Wagner, J. K., Cooper, J. D., Sterling, R., & Royal, C. D. (2012). Tilting at windmills no longer: a data-driven discussion of DTC DNA ancestry tests. *Genetics in Medicine*, *14*(6), 586-593.

Shriver, M. D., & Kittles, R. A. (2004). Genetic ancestry and the search for personalized genetic histories. *Nature Reviews Genetics*, *5*(8), 611-618.

African Ancestry. African Ancestry. (n.d.). Retrieved October 20, 2021, from https://africanancestry.com/.