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Citation	Bolnick, Deborah A., Duana Fullwiley, Jonathan Marks, Susan M. Reverby, Jonathan Kahn, Kimberly Tallbear, Jenny Reardon, et al. 2008. Response to "The legitimacy of genetic ancestry tests." <i>Science</i> 319(5866): 1039-1040.
Published Version	doi:10.1126/science.319.5866.1039a
Accessed	February 17, 2015 6:47:07 PM EST
Citable Link	http://nrs.harvard.edu/urn-3:HUL.InstRepos:3007651
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Response to The Legitimacy of Genetic Ancestry Tests

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Our purpose in writing this policy Forum (19 October 2007, p. 399) was not to persuade the public that tests for genomic ancestry are wholly illegitimate, as Frudakis assumes. Rather, it was to call attention to this influential commercial enterprise and the need for consumers (and the public) to better understand the capabilities and limitations of the available tests. We also hoped to inspire genetics and anthropological associations to discuss these issues with their members.

We did not suggest, as Frudakis states, that there is no connection between genetics and societal interpretations of race. Racial identity is shaped by a variety of factors, including social relationships, life experiences, and biological ancestry. Although DNAPrint's Web site states that race reflects more than genetics, it still (as of 2008) leads consumers to believe that race is inscribed in one's DNA. DNAPrint defines the "biogeographical ancestry" measured by their test as "the biological or genetic component of race" (1), and their underlying model reinforces the archaic racial view that four discrete "parental" populations existed in the past. The assertion that there is some sort of discrete genetic component to race is problematic, and there is no evidence that only four isolated populations existed at any point in the evolutionary history of our species (2, 3). Furthermore, an extensive derivative literature makes it clear that many people think that the AncestryByDNA test identifies their racial makeup (4–7).

Frudakis asserts that our Policy Forum claimed that genomic ancestry panels are rife with biased, non-neutral mutations. We made no such claim. Instead, we noted that some ancestry informative markers (AIMs) involve loci that have undergone selection (8). On the basis of the information provided by DNAPrint Genomics, it is clear that some AIMs are skin pigmentation alleles and others are blood protein alleles involved in malarial resistance (1,4, 9–11). It is therefore important to consider whether these markers measure ancestry alone, or whether they also reflect shared environmental exposures (and thus are not always indicative of shared ancestry). Because the AncestryByDNA test does not differentiate between different evolutionary reasons for shared alleles, the test results may be misleading.

Frudakis then objects to the suggestion that genetic ancestry tests are problematic because they may yield incomplete results due to limited sampling. He is right that perfect databases will never exist, and we agree that companies should quantify the uncertainty and limitations imposed by their data bases. DNAPrint Genomics does calculate the bias, error, and confidence intervals of their estimates, but incomplete geographic sampling creates systematic bias that is difficult to quantify statistically. The sampling of a few, widely dispersed populations for marker selection likely influences the test's results. It is also U.S.-biased because it represents a specifically American racial understanding of human difference. Furthermore, most mitochondrial DNA and Y-chromosome tests do not provide any such statistics, so consumers are often unaware that those tests may yield incomplete or uncertain results.

Frudakis suggests that the problem with the AncestryByDNA test is not that it detects shared ancestry between Native Americans and Eurasians, but that this ancestry is

referred to as “Native American.” We agree. Because the shared alleles predate the divergence of these populations and likely originated in Central Asia, it is misleading to use them as markers of “Native American” ancestry.

Finally, it is clear that scientists who work for companies are not inherently more easily corruptible than academic scientists. That said, there are differences in how academic and commercial products are evaluated. Peer review may be imperfect, but it does require academics to convince experts that their conclusions are supported by the data. Commercial products are not subject to the same system of peer review, and it can be difficult to evaluate conclusions based on proprietary databases. Consequently, conflicts of interest may lead to different outcomes in academia and the business world.