Can DNA 'Witness' Race? : Forensic Uses of An Imperfect Ancestry Testing Technology

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> *An intelligent evaluation of facts is often difficult or impossible without the application of some scientific, technical, or other specialized knowledge. The most common source of this knowledge is the expert witness…* [1]

On August 11, 2004 an African-American man named Derrick Todd Lee was convicted for the first of a series of murder and rape cases in South Louisiana. The life sentence he received would be followed by a death penalty ruling in a second case, which included evidence from several others, just a few months later. Lee had a record of questionable peeping behavior, domestic violence, burglary and assault on his "rap sheet" dating back to his youth. According to various accounts, despite several encounters with the law, Lee fell through the cracks as the local police attempted to compile a profile of the perpetrator of the violent serial deaths of seven women in the Baton Rouge area in the early 2000's.[2]

Lee's eventual convictions were largely based on his Y-chromosome STR DNA profile that matched DNA from samples found on the serial victim's bodies. The rulings were also based on a riveting testimony, and identification of Lee, by a would-be victim who narrowly escaped a rape attempt at his hands. One other identification 'match' in the first of Lee's two trials was based on bloody foot prints left on flooring that matched his construction boots at the scene of one crime, and the assessment of those boots as Lee's by his former girlfriend and his 15-yr-old son.[3] Most of this evidence compiled and marshaled by the defense easily resembles standard forensic details that appear in criminal cases of violent aggression. What is less typical about the ways that the Louisiana police and FBI task force narrowed the focus on Derrick Todd Lee is the series of events preceding the procurement of his DNA though a cheek swab. That is, Lee was the first person in the United States to be identified as a possible suspect by an unconventional DNA analysis tool that racially profiled his DNA left at a crime scene.

The technology that purported to read Lee's race in his DNA is trademarked as *DNAWitness*^{TM}. The name is not accidental. It's inventors at *DNA Print Genomics, Inc.*

want to convey the idea that this technology itself embodies the power of the 'expert witness,' possessing the ability to 'call out' the perpetrator, through literal genotyping 'call-outs' of his specific DNA base pairs. Forensic analysis with *DNAWitness*^{TM} is based on a comparison of a sample of unknown origin with a panel of genetic markers called Ancestry Informative Markers, or AIMs. The basic process of an AIMs analysis consists of a comparative exhibition of varying autosomal coding markers and their relative frequencies in four world populations. The goal of this *specific* iteration of the AIMs test, packaged only for forensics as *DNAWitness*^{TM}, is to *infer* the aggregate of phenotypes associated with any one racial category in the United States. Such an inference is based on the extent to which the anonymous sample expresses allelic variations of markers comprised in a panel that is thought to differ in people from the continents of Africa, Asia, Europe, and (pre-Columbian) America.[4] In the case of the south Louisiana serial killer, *DNAWitness*^{TM} yielded 'ancestry estimates' that the perpetrator's genetic make up was 85% sub-Saharan African and 15% Native American. The Louisiana task force's previous search for a 'Caucasian' male was thereafter deemed to be potentially off the mark. The suspect, as deduced by *DNAWitness*, was most likely a 'lighter skinned black man' as inferred from probabilistic ancestry percentages revealed in the perpetrator's DNA.

In this article I examine the use of *DNAWitness*^{TM} to determine the prospective race of a suspect in order to provide evidence to law enforcement for narrowing a suspect pool. I argue that *DNAWitness*^{TM} falls short of legal and scientific standards for trial admissibility, while it eludes certain legal logics with regard to the use of racial categories in interpreting DNA. *DNAWitness*^{TM} can offer vague profiles in many cases, and has a wide margin of error that too often absorbs what might be understood to be important aspects (i.e. substantial percentages) of ancestral heritage, or of a forensic 'racial profile.' Moreover, this technology's individual ancestry estimates are highly vulnerable to social and political interpretations of phenotype, and may be impossible to accurately interpret with a sufficient degree of objectivity, required of both science and law. It is possible, however, that this test may help to predict a range of skin color phenotypes, as was the case for Lee, since many of the AIMs are skin and hair pigmentation alleles.

The AIMs technology (packaged with different names depending on the market and client) as manufactured by *DNAPrint Genomics* is specifically designed to assess allelic frequency differences of coding DNA, or Single Nucleotide Polymorphisms (SNPs). This is important since markers that the test makers interpret as, 'African', or 'European', for example, are also found in other world populations that differ from the prior continental referent populations used by the company (African, European, Native American and Asian) in name and geographic location. This is to say that differences in ancestry profiles may be due to evolution, gene flow, genetic convergence, or genetic drift. The presentation of *DNAWitness*^{TM} test results demonstrates no attempt to distinguish between these different mechanisms of locus possession in individuals or in groups. Direct and unique ancestry (gene flow) is but one among several mechanisms that might explain shared sequence variation among and between racialized individuals. The simple description of a certain frequency, or set of frequencies, as 'African' ancestry may constitute a false designation of 'racial type,' while, conversely, it *might not*. The fact that there is no gold standard for this technology (a specific proprietary test) should make the legal community pause before lauding its potential success and eventual adoption on a broad basis.

Scientific Merit in Science and Law

From the outset, before evaluating scientific criteria for admissibility in a trial setting, it must be clarified that *DNAWitness*^{TM} has not been used at the trial stage, but rather at the pre-trial stage as *prospective* information for investigating officers. Nonetheless, it is critical to consider the scientific standards for legal admissibility to shed light on the ways in which this technology may actually do harm in the courtroom, since its scientific shortcomings can be easily identified with regard to admissibility rules. Furthermore, holding this technology to accepted legal standards with regard to 'expert' use of science and technology will also allow us to better understand *DNAWitness*^{TM}*'s* problematic role in the legal setting at *any* stage.

Legal precedent would have us focus on three federal cases to determine how scientific merit constitutes the rules for admissibility in a court of law. These are *Daubert v. Merrell Dow Pharmaceuticals, General Electric & Co. v. Joiner,* and *Kumho*

Tire Co., Ltd v. Carmichael. Issues of a) "reliability," b) "scientific validity," and c) whether techniques "can be tested" and "falsified" are of critical concern. As stated in *Daubert v. Merrell Dow,* "scientific methodology today is based on generating hypotheses and testing them to see if they can be falsified; indeed, this methodology is what distinguishes science from other fields of human inquiry."[5] More specifically, a "non-exclusive checklist for trial courts to use in assessing the reliability of scientific expert testimony," provided in *Notes to 702, Federal Rules of Evidence*, include:

> (1) "whether the expert's technique or theory can be challenged in some objective sense, or whether it is instead simply a subjective, conclusory approach that cannot be reasonably assessed for reliability;
> (2) whether the technique or theory has been subject to peer review and publication;
> (3) the known potential rate of error of the technique or theory when applied;
> (4) the existence and maintenance of standards and controls; and
> (5) whether the technique or theory has been generally accepted in the scientific community."[1]

DNAWitness fails to meet this basic checklist on several counts. That these standards were established for the use of scientific evidence in a court of law independent of DNA testing notwithstanding, they nonetheless hold for all scientific evidence. [6] Effective December 1, 2000, several amendments to the federal rules, namely with regard to procedure and methods of reliability, made it clear to both the bench and bar "that an attack on the procedure used to test DNA for evidentiary purposes can be an effective challenge to the weight of any DNA evidence admitted." [7] Thus, presenting genetic results in less than exact and recognized ways could prove detrimental to case arguments.

Historical Background: 'Race' Markers Then and Now

Several of the highly "informative" markers at work in *DNAWitness* have long been the focus of human variation studies in physical anthropology and physiology dating back to mid-20th-century US medicine. Biogenetic studies starting in the 1950's centered on the genetics of blood protein phenotypes that differed in Americans of 'African' descent and of those of 'European stock.' During this same time period, vocal social scientists of the day issued statements warning against using such traits to emphasize the

(superficial) nature of phenotypes, given the breadth of the human species' biological commonality. The most noteworthy of such public discussions on the issue was the 1950 UNESCO *Statement on Race*. Written largely by social scientists, yet reviewed by leading geneticists, the authors centered on the "temporary" nature of "varying manifestations" of traits. Their goal was to unsettle the common belief in racial biological determinism and fixity:

> In looking at the different "races" of mankind today…the varying manifestations of physical traits which they exhibit are not "end results" but bills of exchange, as it were, drawn on the bank of time, negotiable securities which can be turned into the coin of any realm with which it is sought to have biological relations. In other words, we perceive the consequences of different histories of biological experiences in the "races" of today. [8]

Still many researchers continued to examine such "end results"—as ends in and of themselves—that "varyingly manifested" in blacks and whites. Nonetheless, even in the 1960s, some scientists working with such physical traits (that today are characterized as some of the "most informative" of the "Ancestry Informative Markers" operative in *DNAWitness*) provided clear caveats against assumptions that the frequent possession of certain alleles in a referent group and in a comparative sample necessarily indicated *direct* and *unique* ancestry *from* the referent group *to* the sample. In fact, in order to draw such a conclusion, ~~or~~ of gene flow, one 1960's researcher wrote that a number of facts needed to be held constant to infer ancestry—facts with regard to the *certainty* of a population's putatively fixed characteristics, both genetic and ethnic. Zoologist, anthropologist and pediatrician T. Edward Reed, who provided a detailed review of 11 studies on such markers as of 1969, warned that researchers had not "always appreciated" the simple and obvious criteria that must be met in order to estimate ancestry from what are now Ancestry Informative Markers.

He wrote:

> Critical evaluation of estimates of M [Caucasian mixture in 'American Negroes'] requires complete specification of the needed criteria and judgment on the degree to which these criteria are met. These criteria are simple and obvious, but the demands they make have not always been appreciated. They are as follows:

1) The exact ethnic compositions of the two ancestral populations, African Negro and Caucasian, are known;

2) No change in gene frequency (for the gene in question) between ancestral and modern populations either of African Negroes or of American Caucasians has occurred;

3) Interbreeding of the two ancestral populations is the *only* factor affecting gene frequency in U.S. Negroes –that is, there has been no selection, mutation, or genetic drift (Emphasis mine). [9]

As stated in the introductory section, "admixture" analyses (based on the use of several of these same markers reviewed by Reed) now used today in *DNAPrint*'s products specifically do *not* take into account these other possible sources of varied gene frequency distributions in present-day populations enumerated nearly 40 years ago. Other researchers in the 21st century have repeated Reed's call. [10] Yet, as concerns *DNAPrint's* various AIMs products, these points have gone overlooked. Instead, the company has pursued this technology by creating a methodology that is seemingly robust if allowed to operate independent of any recognition of this larger set of criteria.

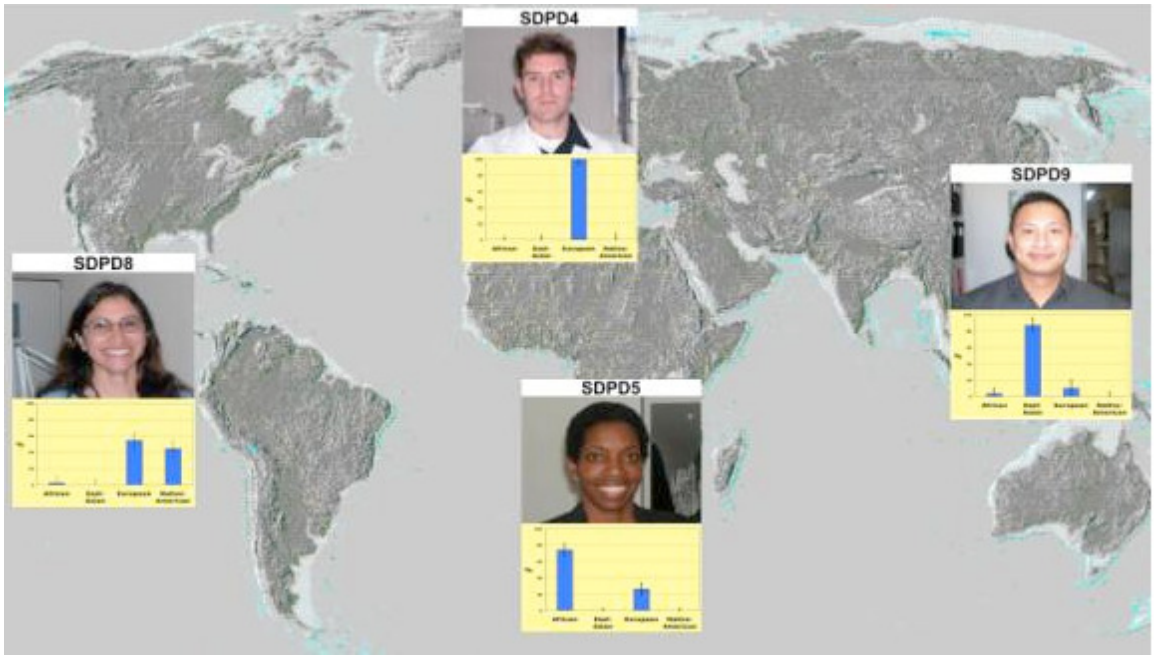

Figure 1. Sample volunteers who sent in DNA and photos to be processed for *DNAPrint*'s forensic database. They each possess an ID number, while their estimated continental ancestry is listed on a bar graph below their photos. The photos are placed on the

"continent of origin" that *DNAPrint* scientists believe best represents each individual. (Unaltered image from the website for *DNAWitness*).

When compiling previous studies of unevenly distributed gene frequencies of a handful of known traits in Africans, East Asians, Europeans, and Native Americans, *DNA Print* scientists initially identified the continental groups of interest, and then examined human DNA samples from public genetic databases, such as *dbSNP*, to identify 34 genetic markers that displayed at least a 50% allelic frequency distribution difference between any two of the four groups. To determine this, all subjects in a defined group, for example 'Africans', were tested for the panel of alleles. Then, the researchers determined the frequency at which AIMs appeared in each group as a whole. These referent populations surely displayed as much genetic heterogeneity as any group, but the point of the AIMs research was to find those specific alleles that they shared at a certain frequency, as long as that frequency, when contrasted with at least one other of the four groups, demonstrated a difference of 50%. Such purified, or *artificially* homogenized, samples came to serve as 'parental populations' of Africans, Europeans, Native Americans, and East Asians for 'contemporary' test takers.

AIMs analyses most often rely on a series of unknowns, which are the points 1-3 that Reed highlighted as necessary to proceeding with confidence with this model. When researchers cannot answer questions, such as 'the exact ethnic composition' of the contributing parental source populations, or even how many sources there were in certain terms, they instead rely on assumptions. These assumptions are not arbitrary. They are usually informed by a given society's historical account of when the two or more groups who are the putative 'parentals' encountered each other. But problems arise when social context and historical accounts meet with sampling limits. For instance, using contemporary acknowledged Native American groups as one source population for Mexican Americans might overlook an actual history of genocide of peoples who no doubt contributed to present-day populations in the Americas, but who may no longer exist. [10] To complicate matters more, Mexican Americans often have more Amerindian heritage than the referent groups who are posited as their Native American ancestors. For political and historical reasons Native Americans need only possess 1/8 (12.5%)

demonstrable Native American ancestry, whereas Mexicans may have considerably more. [11] Finally, in addition to these quandaries, when alleles that have a high frequency in the specific reference groups tested (those labeled African, European, Native American, etc.) appear in a "client" taking the test, the AIMs test reads that the client, himself or herself, has inherited the *specific referent ancestry* rather than say ancestry (or SNPs) from *other* still un-sampled parts of the globe.

We may now revisit the *Federal Rules of Evidence* "checklist" for scientific admissibility in the legal sphere. It should be clear that the basis on which *DNAPrint's* "parental populations" are constructed, and artificially homogenized, involves a certain cultural bias, thus subjective choice, of racial typing from the outset. This is a critique that has been levied against *DNAPrint* recently, and which highlights the *systematic* bias of the results its technologies yield. [12,13] Although various studies that have used these markers have undergone peer review, the scientific journals and the professional peers of *DNAPrint* scientists and their collaborators have not requested that the entire set of markers, the complete details of sampling, and the assumptions that populations' trait possessions are due to gene flow, be evaluated. Thus, the full scope of this technology remains proprietary, and evades peer review. The technology continues to provoke controversy due to its modeling of four archaic racial types and its claim to be able to estimate individual ancestry percentages from these types without any discussion of other mechanisms for allelic similarity. [12-14] Additionally, although various AIMs products' margins of error may be known, the company website lists different, smaller, figures (ranging from .5 to 15%) than its sole scientific advisor has admitted to publicly (as high as 30 %). [15, 16]. On several counts, *DNAWitness* fails to meet items 1-5 of the "checklist."

Conclusion

The rise of new genetic technologies in the past two decades has yielded a range of scientific possibilities for the courts. Not all genetic tests perform the same kinds of tasks, and none were instituted without prolonged discussion, debate, and research consensus with regard to their reliability and consistency among scientists and law

enforcement. [17] As this analysis makes clear, *DNAWitness* is based on Ancestry Informative Marker technology, or coding SNPs, that are largely shared among individuals and groups for varying reasons—reasons that are neither described nor acknowledged explicitly in the test results offered by *DNAPrint*. AIMs-based technologies, like *DNAWitness*, are attempts to model human history from a specifically American perspective to *infer* present-day humans' continental origins. [18] Such inferences are based on the extent to which any subject or sample shares a panel of alleles (or variants of alleles) that code for genomic function, such as malaria resistance, UV protection, lactase digestion, skin pigmentation, etc. There is a range of such traits that are conserved in, and shared between, different peoples and populations around the globe for evolutionary, adaptive, migratory, and cultural reasons. To assume that people who share, or rather *co-possess*, these traits can necessarily be 'diagnosed' with a specific source ancestry is misleading. Not only will siblings often share the same profile, *or not*, but individuals from all four 'parental' continental groups offered up by the model could feasibly share similar profiles, *or not*. As a forensics market version of the AIMs technology, *DNAWitness* may offer precise mathematical ancestry percentages, but the accuracy of that precision remains debatable.

At best, this technology is an experimental modeling tool that hopes to mimic recent American human history as it reconstructs four racial types through an artificial homogenizing of markers found with relatively higher frequencies on some continents and lower frequencies on others. As compelling as *DNAWitness* as a tool may seem, investigators should require that DNA analyses used in the serious proceedings of law be falsifiable, reliable, and thoroughly vetted. Anything less would prove irresponsible if incorporated into criminal investigations.

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