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Molecular anthropology: the judicial use of genetic data in archaeology

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A B S T R A C T

Anthropology has always been an unashamed scavenger discipline, acquiring and employing techniques developed in other physical, life and social sciences to apply to a holistic approach to studying humanity. In this regard, the adoption of genetic analyses into archaeological investigations has paralleled many previous adoptions including those of radiometric dating, stable isotope analysis and chemical analysis of material culture. Employing DNA data in reconstructions of prehistory, however, has been hampered particularly by the expense of generating the data – both financial and logistical – and, at least initially, by unwarranted resistance to take seriously molecular data. While the expense continues to rise as new techniques become available, there has been a reversal in the place of genetic data in that it is now privileged over other sources of data. This kind of molecular chauvinism leads to overreach in interpretation and is no less likely to hamper our progress. Moving forward we would do best be judicial in the use of genetic data alongside other independent archaeological evidence in reconstructing the past.

1. Introduction

Modern and ancient DNA data are now routinely incorporated in reconstructions of the past. Ancient DNA (aDNA) loosely refers to any DNA that has degraded. The techniques employed in the analysis of aDNA are applied both to specimens of tremendous antiquity (e.g., Orlando et al., 2013) and those dead only a century (e.g. Miller et al., 2009). The DNA itself, as a consequence of its poor state of preservation, is both low quality and low quantity. Nonetheless, with ever improving technologies the field has gone from sequencing tiny fragments of DNA from museum specimens of preserved muscle tissue (Higuchi et al., 1984), to complete or nearly complete genomes of extinct human species (Briggs et al., 2009; Green et al., 2010; Lalueza-Fox et al., 2011; Reich et al., 2011; Reich et al., 2010). Just as there have been leaps forward in the technical ability to generate molecular data that is of interest to archaeology, there has also been a sea change in how it is treated within the field. In the late 1960s, the results of molecular anthropological studies were met with resistance and mistrust. Today, molecular data in general, and ancient DNA data in particular, have been warmly embraced by prehistorians. The purpose of this brief review is not to use the benefit of hindsight to admonish archaeology's resistance to genetic data, but to highlight the growing problem of overreach. Genetic data is often placed above all other evidence, which is equally troubling, where a judicious use of molecular datasets would be more appropriate.

2. Anthropology's fraught relationship with molecular data

The incursion of molecular data into anthropology began in earnest when Vince Sarich and Allan Wilson published their immunologically informed phylogeny of the apes, including humans (Sarich and Wilson, 1967). Most startling to the paleoanthropological community was their conclusion that the common ancestor of humans and the other African apes lived about 5 million years ago. Most paleoanthropologists in the 1960s accepted 30 million years ago as a reasonable estimate of the age of the common ancestor (Pilbeam and Simons, 1965). So enormous a discrepancy meant that no one could see a way to reconciling the two reconstructions. Sides were taken. Heels were dug in.

Much of the vitriol surrounding the issue was voiced at conferences, but plenty made it into print. Sarich and Wilson were painted as outsiders without sufficient respect for the morphological data, with Simons (1968:328) carefully distinguishing them

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from those who had done the hard work of studying the fossil record appropriately:

> Students of human origins will know, however, that the story of hominid[1] origins begins much earlier than this, since hominids of the genus *Ramapithecus* date back to the late Miocene, about 14 million years ago.

While Sarich and Wilson, though, were being chastised for their failure to appreciate the fossil record, many paleoanthropologists felt comfortable dismissing data they did not understand. Louis Leakey (1970:746–7), for example in 1970, wrote "I am not qualified to discuss the biochemical evidence…" but that "[t]he date of separation suggested by Wilson and Sarich, i.e., only five million years ago, is not in accord with the facts available today."

With the benefit of more than forty years accumulated history, it is too easy to paint Sarich and Wilson as the heroes, and the paleoanthropologists as the narrow-minded villains whose recalcitrance delayed advances in the field. Sarich and Wilson could, though, have done more to explain their methods and results. Sherwood Washburn, who had been Sarich's PhD advisor urged him to write a paper that would be accessible to the general anthropological community. Expressing frustration that such a paper would be necessary, Sarich responded, "That's all published. People should read what's published, and they should accept it" (described in Lewin, 1987). More recently, many molecular anthropologists have made considerable efforts to communicate with anthropologists not trained in molecular biology (e.g. Brown and Brown, 1992, 2011; Kaestle and Horsburgh, 2002; Matisoo-Smith and Horsburgh, 2012; Mulligan, 2006; O'Rourke et al., 1996, 2000), but none of these came soon enough to help the paleoanthropologists working 1960s and 1970s comprehend the new data and integrate them into their models of human evolution.

It took longer than it needed to, but paleoanthropologists did eventually come around and began integrating the molecular and fossil data. Slowly the dates attributed to the common ancestor of all the great apes slid away from 30 million years ago and closer to 15 million years ago. By 1984 David Pilbeam wrote that, "[t]he earlier debate between physical anthropologists and molecular biologists over the pattern and timing of hominoid evolution is now basically settled" and even self-deprecatingly asked "Why was the hominoid fossil record misinterpreted by dimmer paleontologists such as me?" He concluded that fragmentary fossil remains had been relied upon too heavily in the reconstruction of phylogenetic relationships; that the Miocene apes are taxonomically more diverse than are the extant apes as well as more morphologically heterogeneous rendering difficult the task of discerning relationships among the extinct species, and between the extinct and extant species.

3. The overcorrection

By the mid-1980s the resentment felt by paleoanthropologists at the infiltration of their field by biochemists, geneticists and molecular anthropologists had waned, and most embraced the news lines of evidence as valuable contributions to the greater mission: the development of accurate, precise and rich explanatory models of human evolution. Where molecular data were once disregarded, we have now swung too far in the other direction, and people studying a human evolution from a genetic perspective have become as chauvinistic about their data as the paleoanthropologists ever were. I highlight a single quote here, but contend that the authors are not alone in their perspective:

> The best way to understand our evolutionary history as modern humans is comparing our own genome with those of our closest relatives. The genetic bases of the traits that we do not share with them are going to be those that define our singularity as a species (Sánchez-Quinto and Lalueza-Fox, 2015).

There are three fundamental problems with such a position: 1) DNA is not a blueprint for an organism; 2) important developments in human prehistory need not have been universally rooted in genetic change; and, 3) privileging the genetic data over all the other classes of data available impoverishes the nature of the reconstructions available to us.

3.1. The relationships between genotype and phenotype

The metaphor of DNA as a 'blueprint' or 'program' is an attractive and seductive one. Blueprints share a one to one correspondence with the object they specify; they always produce the same results. This is certainly not the case with DNA. Marks (1996) has described this perception of genetics as "high tech astrology" (p6) with genes being viewed as 'predisposers' in some sort of soft determinism. We are pretty good at looking at DNA and telling you if someone was lactose tolerant, or had sickle cell trait, or were bitter (PTC) tasters. We are terrible at looking at DNA and telling you if someone was musical, short-tempered, introverted, athletic, creative – that is, we are terrible at telling you most of the things that are likely to be of interest. The heritability of human height has long been the subject of research interest. As multiple studies make clear (Aulchenko et al., 2009; Gudbjartsson et al., 2008; Lettre et al., 2008; Visscher, 2008; Weedon et al., 2008), despite genome wide association studies (GWAS) in thousands of people, attempts to locate genetic variants strongly associated with variation in human height have proven underwhelming. Somewhat more than 50 variants have been identified that are associated with variation in human height. Combined, however, the variants can account for only 4–6% of the measured variation in human height. As Aulchenko et al. (2009) point out, if you want to predict the height of a person, you are currently better off employing the method that Galton published in 1886 (involving little more than averaging the heights of both parents and correcting for sex) than you are with a genome-wide survey of variation.

Other characteristics of importance are likewise poorly explained by existing surveys of genetic variants. A GWAS study attempting to located genes involved in the development of facial morphology was able to implicate five candidate genes (Liu et al., 2012). Just as in the studies of human height, however, the discovered variants contributed very little to the variation in human facial morphology. More recently an attempt to use genetic variation to calculate a predicted facial morphology (Claes et al., 2014). The study has been criticized on statistical grounds (Hallgrimsson et al., 2014). No correction was made to account for multiple comparisons, and further analyses showed that only one of the original 46 candidate genes was sufficiently significant to survive Bonferroni correction.

A final example I will offer here involves a GWAS in search of the genetic underpinning of personality traits. In the 1980s Cloninger

[1] This discussion centers on the 1960s and 1970s, and so hominid is used to refer to humans, their ancestors and their closely related species since the divergence of our lineage from that of chimpanzees and bonobos. This older taxonomic scheme placed chimpanzees, gorillas and orangutans in the family Pongidae, and humans in the family Hominidae (hence, hominid). The taxonomy was revised to take seriously the notion that a taxon including chimpanzees, gorillas and orangutans but excluding humans is one constructed without reference to the biological reality that chimpanzees are considerably more closely related to humans than they are to gorillas and orangutans. See Wood and Richmond (2000).