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Catherine Bliss

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Mapping Race through Admixture

Catherine Bliss, The New School for Social Research, New York, UNITED STATES

Abstract: Mapping Admixture Linkage Disequilibrium (MALD) is a technology that separates genomic ancestral lineages to identify disease genes. In the U.S., where a significant segment of the population has unknown ancestral origins, researchers use MALD to tease out continental haplotypes and (re)assign ancestry to disease samples. While MALD is fast-becoming a primary medical genetic technology, its publicly known uses lie in the service fields of recreational DNA genealogy and forensic profiling. Here, private companies use MALD to tell clients where their ancestors likely came from or to advise law enforcement on what kind of racially-defined features to look for in a suspect. This paper looks at the practical assemblage of MALD applications and its effects in defining ancestry in terms of race. Through this assemblage, society produces the genome as racial and race as genetic. Moreover, identity is refashioned through a genomic knowledge of self.

Keywords: Knowledge, Genetics, Race

Introduction

MAPPING BY ADMIXTURE Linkage Disequilibrium (MALD) is a rising genomic technology that maps ancestral lineages onto a continental matrix in order to assist in the hunt for disease genes and human origins. When MALD was invented in the medical field of the early 1990s, it emerged from a context wherein "race" was the operative framework for understanding human difference. MALD has now been applied in the private sectors of forensic genetics and genetic genealogy to translate ancestral data into socially useful information. In this consumer arena, MALD continues to intersect with concepts of "race" circulating in the wider society. As a result, both MALD practices and common sense norms about "race" have changed.

Here, I discuss the dialectic of MALD technology and racial ideas by analyzing its practices in different fields: medicine, forensic genetics, and genetic genealogy. Using evidence from scientific articles, news media publications, and field notes of meetings with leading MALD researchers, I argue that the practices and practical networks that fostered the development of MALD could only happen in the presence of shared and legitimized values regarding human difference. To this end, I explore the broader context of social norms that condition these fields.[1]

Instrumental to my task are the concepts of *practice* and *assemblage* delineated by Paul Rabinow. In recent studies of biotechnology, Rabinow attempts to illuminate the contours of collaboration between "distinctive *subjects,* the *site* in which they worked, and the *object* they invented" (Rabinow 1996:2). He defines *practice* as "norms in context and in process" (Rabinow 1996:14). This definition suggests that social action and norms cannot be separated. Social values that make certain actions sensible, justifiable, or just plain thinkable are embedded in the choices that agents make. Hence, following the rise of a new technological practice involves tracking the norms or values that imbue that very process.

Rabinow's notion of *assemblage* highlights a certain emergent moment of collective practice. He states:

> [Assemblages] are a distinctive type of experimental matrix of heterogeneous elements, techniques, and concepts. They are not yet an experimental system in which controlled variation can be produced, measured, and observed. (Rabinow 2003:56)

Because the relationships between institutional domains in an assemblage are new, they are more flexible and impressionable with regards to norms circulating in the greater society. Here, I apply this concept to the emergent alliances between medicine, forensics, and genealogy that have brought MALD to bear.

Early Inklings

MALD is often presented as a technology *de novo* – a technology filling a hole only made obvious by the molecular turn in genetics. However, the idea of MALD has been brewing in population genetic circles for quite some time. The theory of MALD was, indeed, devised in the 1950s. American population geneticists facing the glaring evidence of post-

[1] For an analysis of the use of "race" in a contemporary MALD research setting, see Fullwiley 2007a and 2007b.

slavery miscegenation argued that "American Negroes" were like a cross-bred population in a living laboratory. Outlining the theory's most basic tenets, MALD inventor David C. Rife stated:

> A correlation in the occurrence of two genetic traits within a population is usually interpreted as evidence against, rather than for linkage. Linkage results in correlations within families, but in opposite types of correlation from one family to another...opposite types of correlation within different families cancel correlations within the population as a whole…Relatively new populations of hybrid origin provide an exception to the foregoing rule. Under specific circumstances correlations within them may be indicative of linkage. (Rife 1954:26)

Rife argued if only geneticists could hold elements of the parental hereditary material constant, other hereditary material would become visible. What geneticists needed to find were traits certainly constitutive of differentiated populations.

Rife worked with traits then supposed to be hereditary: blood groups, pigmentation, surface characteristics, and taste sensitivity. Unconvinced of the environment's role in shaping these traits, Rife went so far as to argue for applying his theory to a study of socially relevant traits such as intelligence. On the one hand, Rife's biological scrutiny of admixture countered a long history of discourse that barred racial admixture as naturally and socially abominable – a discourse responsible for generations of antimiscegenation legislation. On the other hand, Rife's approach came at a time when social and cultural explanations for "race" were rapidly replacing genetic or biological explanations in the public. While Rife's basic argument would later be retrieved and taken up by population geneticists working at the molecular level, in the wake of the American Eugenics Movement and the recently revealed atrocities of Nazi *Rassenhygiene*, Rife's MALD went undeveloped and unexplored.

A New Life

After lying dormant for over thirty years, a team of scientists in two American laboratories reopened dialogue about using admixture to understand human heredity. The first laboratory to revisit Rife's theory was the University of Texas Health Science Center's Center for Demographic and Population Genetics. There, Ranajit Chakraborty, a researcher heavily involved in establishing forensic genetics, sought a shortcut for candidate gene hunts. He hoped the theory of MALD would make the African American and growing Latino American populations of the US analyzable for forensic and medical researchers (Chakraborty 1986). The Center's J. Claiborne Stephens was also interested in finding this shortcut. When he transferred to the Laboratory of Viral Carcinogenesis at the National Cancer Institute, Stephens co-penned the first MALD articles that included computerized tests (Stephens, Briscoe and O'Brien 1994; Briscoe, Stephens, and O'Brien 1994). Under the wing of these seasoned scientists, MALD took off as a federally sponsored research method.

At the National Cancer Institute, MALD was first applied in human disease study and animal conservation efforts. Laboratory Chief Stephen J. O'Brien had a long history of solving population decimation issues with newly emerging molecular technologies. O'Brien's laboratory was at the center of high profile discoveries surrounding feline leukemia and population resistance to HIV-1. At this site, MALD coevolved with several research frameworks: disease discovery, conservation by ancestral analysis, and population identification.

In all of the initial publications on MALD, the term "race" is explicitly applied (Chakraborty and Weiss 1986; Stephens, Briscoe and O'Brien 1994; Briscoe, Stephens, and O'Brien 1994). These researchers believed that populations on continents had been long enough isolated to produce "genetically differentiated racial groups" (Stephens, Briscoe and O'Brien 1994), but, what's more, they believed that in studying admixed populations – populations that hold the status of "racial minority" in the US – they were bringing genetics to groups who would otherwise miss out (fieldnotes Aug 8, 2007). In the wider social arena, this discourse of "serving the racially underserved" was quickly gaining ground. In social scientific circles, scholars and policy analysts increasingly spoke on the "racial divide" in US living conditions (see Haney-Lopez 1995: 546; Ong 1999, Omi and Winant 1994). They recommended that officials take a proactive role in redressing the legacy of discrimination. To this end, social scientists worked with federal representatives to perfect a standardized racial category scheme for federally funded programs. Likewise, the federal government launched several health disparities research campaigns focused on minority health (see, for example, HHS 1994 and NIH 1993). These campaigns pivoted on the deployment of the federal "race" scheme crafted by the federal government. Additionally, the National Institutes of Health founded two in-house minority research offices: the Office of Women's Health Research and the National Center on Minority Health and Health Disparities. Thus, the practice of using "race" in MALD medical research reflects widely held social norms of the time.

Growing Momentum

Over the next ten years, the development of MALD was to pick up speed in the hands of students emerging from these laboratories. One of Chakraborty's student in particular, Mark Shriver, has brought MALD to the public while remaining firmly invested in medical and evolutionary research. Shriver developed his doctoral studies interest in forensic markers to a full-fledged search for MALD markers that could be used in both the forensic and medical context. By the late 1990s, Shriver's new laboratory at Pennsylvania State University was responsible for the release of the first MALD marker panels (Shriver et al. 1997; Parra et al. 1998). These panels fulfilled Rife's early dream: they contained ancestry informative markers that would allow any geneticist to hold genetic traits in constant, so that hard to distinguish underlying traits would stand out. During this time, Shriver's laboratory was funded by both the National Institutes of Health (NIH) and the National Institutes of Justice (NIJ). The assemblage of institutional interests first established by the co-invention of MALD in the forensic-centric Center for Demographic and Population Genetics and the public health-focused National Cancer Institute further sedimented with Shriver's collaborative laboratory. This laboratory became a hotspot for normative decision-making in the world of MALD research.

MALD also gained ground in the laboratory of a doctoral student working with O'Brien. David Reich of Harvard Medical School developed MALD into a technology that uses the population frequencies of thousands of markers to bring disease genes to light. While Reich chooses to focus on medical applications of MALD, resisting collaboration with forensic programs, his position as an associate of Harvard & MIT's Broad Institute has also widened the scope of MALD's institutional assemblage (fieldnotes Oct 3, 2007). Broad's esteemed relationship with the NIH's National Human Genome Research Institute – a relationship forged during the Human Genome Project and International HapMap Project – has ensured top dollar will be spent on new applications of MALD. Thus, through the legacy these students have promoted, the two federal health institutes responsible for funding the majority of genomic research in the US – the National Cancer Institute and the National Human Genome Research Institute – have further committed their support of the operationalization of MALD. In turn, these laboratories remain beholden to federally supported social norms about "race" and human difference such as the expansion and amendment of federal "race" categories and the acceptance of multiraciality as ushered in with the 2000 Census.

MALD in the Private Sector

Intimations of the kind of DNA imaging and origins information that MALD technologies now make possible have long circulated in scientific fiction and popular cinema. In turn, MALD has enjoyed a rapid entry into the private sector. Much of MALD's consumer relevance has grown out of the development of one statistical program that has been released in the forensic and genealogy markets. Shriver developed an algorithm that analyzes 176 ancestry informative markers for their continental frequency and uses the composite percentage to determine the "admixture proportion" of an individual sample. Shriver co-owns the rights for this program with a private company called DNAPrint Genomics. DNAPrint Genomics was already in the business of paternity testing and forensic service, but the company first gained public recognition for itself and for MALD when they applied this algorithm to create physical profiles based on DNA evidence. The success with one long unsolved case in particular, the case of the Louisiana Serial Killer Derrick Todd Lee, gained DNAPrint Genomics popular acclaim. In this case, as in others, the company's admixture breakdown led law enforcement officials to reverse their notion of the "race" of a suspect. Likewise, in the genealogy realm, this algorithm has made a stamp on the public consciousness with its unique origins reporting system. DNAPrint provides a diagram of admixture proportions that maps individual ancestry as a spectral point amidst a continental matrix easily translated into racial terms.

However, with these inroads into the forensic and genealogy domains, new practices have emerged. In the forensic realm, MALD initially enmeshed with common sense racial schemes, such as those regularly employed in law enforcement. Law enforcement officials used admixture analysis to assign a racial value to suspects. For example, in the aforementioned case of the Louisiana Serial Killer, officials believed the suspect was a "white" male. However, the admixture analysis indicated the suspect had mostly (in DNAPrint Genomic's terms) SubSaharan African and Native American descent. This led officials to search for a "black" male instead (Simons 2003). Similarly, in a parental homicide of a four-year-old girl, tests indicated that police should search for a "racially mixed" couple (Willing 2005). The ascertained admixture of the victim led investigators to code their suspects as "white" and "black" (Simons 2003).

Yet, as law enforcement routinely takes criticism for using "race" to catch criminals, forensic genetics has also felt the public sting of using "race"-based practices. As a result, companies like DNAPrint have attempted to distance themselves from common sense constructions of "race" even as customers use their

product with "race" in mind. Note a remark by Shriver at the height of case-solving success: "By showing the continuum of genetic variation among people, our test dispels race as a scientific way of categorizing people" (Willing 2005). While companies claim that they cannot be responsible for the way law enforcement uses their service, they attempt to assuage the public that they do not buy into clear-cut racial category schemes (fieldnotes October 5, 2007).

This normative change has, likewise, changed the way MALD is practiced and presented in the genealogy and medical sectors. While genealogy companies often early on made claims to determine customers' racial origins, today companies use terms like "ethnicity" and "tribe" to elaborate promises to customers. Public and private laboratories have begun straying from the continent-based matrix that so easily maps onto a racial framework, using MALD to tease out finer ethnic origins (see Campbell et al. 2005; Seldin et al. 2006; Helgason et al. 2005; Choudhry et al. 2006; Parra et al. 2001; Kim et al. 2005; Listman et al. 2007). Many leaders in the field are adamantly pushing for a complete move away from continental stratification (fieldnotes October 5, 2007). As noted, leaders in one field are more often than not responsible for the success of MALD in all fields. Thus, small analytical shifts have a domino effect. DNAPrint, for example, now offers a sub-European ethnic breakdown in their forensic and genealogy platforms. The frontier-like bond between medicine, forensics, and genealogy permits an easy cross-pollination and incorporation of broader social norms.

Conclusion: Rescripting "Race"

The assemblage of medical, forensic, and genealogy MALD shown here allows us to witness the dialectical relationship between practices and norms in modern day technology. MALD emerged from a context where certain racial ideas held sway and its practices have been changing with respect to shifting racial ideas ever since.

In the case of recent alterations to MALD practice, we can see that public disapproval of racial profiling in law enforcement and medicine marks a recent change in social norms about "race" in general. In the late nineties, the concept of "race" received much critical attention both academically and publicly. Scholars questioned the fixity of racial identities over time (Hall 1996; Loveman 1999). Affirmative Action programs were challenged in the courts and, in many states, scaled back. Federal standardizations came under attack as a small group of multiethnic activists raised the issue of multiple ancestral origins. The culmination of these efforts can be witnessed in the Census policy initiated in 2000 that permits US residents to mark more than one racial category. This isn't to say "race" has been ousted from legitimate scientific practice. The debate over what "race" means that continues to rage in medical genomics can attest to the unresolved nature of knowledge about "race" in society.

This conundrum has serious consequences for individual subjectivity. Following MALD's entry into the greater public consciousness, the laity has responded with new ways of conceptualizing human origins, personal identity and the self. Documentaries elaborating the concepts of ancestry and admixture have been sponsored by major media organizations (Journey of Man Motherland 2004; African American Lives 2006). In primetime television and news programs, genetic genealogy testing is becoming a well-known way to create personal narratives (Bashir 2007; Daly 2005; Hawks 2006). In college admissions and citizenship claims, MALD is affecting how institutions distribute resources and determine admission (Tanner 2005; Harmon 2006). MALD is increasingly serving as a fertile ground for ideas about who we are and how genetics matters for our public persona. It will be interesting to see how the precipitation of new ideas about "race" is altered by and alters genomic technology in the coming years. Likewise, it will be worth watching how the instantiation of MALD in new fields and the sedimentation of relations between older fields affect the dynamics of normativity in MALD research across the board.

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About the Author

Catherine Bliss
Catherine Bliss is a doctoral candidate at the Department of Sociology of the New School for Social Research. Ms. Bliss has an MA in Sociology and an MA in Ethnic Studies. Her research interests are scientific knowledge production, cultural studies of scientific communities, and social constructions of race. She is currently completing a dissertation on molecular genetic productions of race and ethnicity.

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