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Ambiguity and Scientific Authority: Population Classification in Genomic Science

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Abstract

The molecularization of race thesis suggests geneticists are gaining greater authority to define human populations and differences, and they are doing so by increasingly defining them in terms of U.S. racial categories. Using a mixed methodology of a content analysis of articles published in *Nature Genetics* (in 1993, 2001, and 2009) and interviews, we explore geneticists' population labeling practices. Geneticists use eight classification systems that follow racial, geographic, and ethnic logics of definition. We find limited support for racialization of classification. Use of quasi-racial "continental" terms has grown over time, but more surprising is the persistent and indiscriminate blending of classification schemes at the field level, the article level, and within-population labels. This blending has led the practical definition of "population" to become more ambiguous rather than standardized over time. Classificatory ambiguity serves several functions: it helps geneticists negotiate collaborations among researchers with competing demands, resist bureaucratic oversight, and build accountability with study populations. Far from being dysfunctional, we show the ambiguity of population definition is linked to geneticists' efforts to build scientific authority. Our findings revise the long-standing theoretical link between scientific authority and standardization and social order. We find that scientific ambiguity can function to produce scientific authority.

Keywords

genetics, race, ethnicity, geography, standardization, ambiguity, authority

One of the most commented upon puzzles of the postgenomic era is the paradox of race. When President Clinton held a press conference on June 26, 2000, to announce the completion of the first draft of the sequence of the human genome, the main message he and scientific leaders Francis Collins and Craig Venter chose to emphasize was that of universal human similarity: "in genetic terms, all human beings, regardless of race, are more than 99.9 percent the same" (quoted in Bliss 2012:1). Venter, Collins, and other leading scientists strongly stated that contemporary

genetics shows race not to be a scientific concept, and that "precise racial boundaries" cannot be legitimated scientifically. Yet this apparent consensus was soon broken. Geneticists

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Neil Risch, Noah Rosenberg, and others argued that genetic data could map global genetic variation in ways that closely approximate common sense ideas of race, that genetic data could accurately assign most individuals to a racial group, and that racial differences would be important to account for in genetic studies of diseases (Risch et al. 2002; Rosenberg et al. 2002). Debate continues to rage about these various claims and also whether race is indeed a genetically valid concept, and thus whether race can legitimately inform the application of genetics to medicine, pharmacology, and forensics.

The postgenomic resurgence of racial debate has spawned two lines of social science commentary that are in tension with each other. The first notes the simultaneous racialization of biomedical science and the molecularization of race. Race-based knowledge has gained salience in genetics and biomedicine, due to movements to foster greater inclusion of women and minorities in medical research (Epstein 2007), scientists' own justice-oriented political commitments (Bliss 2012; Fullwiley 2008), and markets for race-targeted genealogical and pharmaceutical products (Kahn 2012; Pollock 2012). The race concept is being reconceived in molecular terms, and again appearing as a biological human essence, even as geneticists reframe racial classification in terms of varying biogeographic ancestry and admixture (Duster 2006; Fujimura and Rajagopalan 2011; Gannett 2014). And even neutrally presented genetic findings encourage essentialist understandings of race among the lay public (Phelan, Link, and Feldman 2013).

While the first line of research emphasizes the biogenetic "hardening" of race, a second, less prominent line emphasizes conceptual incoherence, or "softening," in the ways geneticists and biomedical researchers define race and classify populations in their studies. As Jonathan Marks (2012) has said,

Geneticists tend to take their populations as 'natural' units in a way that troubles anthropologists. That is why you can find a

linguistic group (such as Bantu) contrasted against a political state (French) and an ethnic label (Druze) in the same genetic study, without any awareness that they refer to entirely different kinds of belonging. When geneticists have queried the construction of their samples, they tend to place an uncomfortable emphasis on 'purity of ancestry', denying the realities of human history, with all its murky interbreeding and complexity. Looking for admixture, after all, presupposes a primordial state without it.

Marks notes that genetics researchers seem to uncritically compare population categories that are apples and oranges and make unwarranted assumptions about the origins and composition of those populations. Studies of geneticists note that when asked plainly to define what they mean by "race," geneticists often find themselves surprised to be unable to provide a clear answer (Bliss 2012; Fujimura and Rajagopalan 2011; Fullwiley 2008; Lee et al. 2008).

Both perspectives are mobilized to criticize the potential reification of race, the former from the perspective of essentialism, and the latter from the fear that lack of a coherent concept of race will enable folk-thinking about racial purity, essentialism, and difference to persist. However, the critical attention to racial reification in genetics has led to the underappreciation of two issues upon which this article focuses. First, the focus on race has diverted attention from the diversity of ways that geneticists classify human populations. Second, the concern with reification has led to the neglect of the symbolic functions of the incoherence of geneticists' classifications.

This article explores the ambiguous and flexible ways that geneticists classify human populations and differences. We document the scope of classificatory ambiguity with a content analysis of articles from *Nature Genetics*, the field's leading journal. We find that (1) over time, population labeling has increased to near ubiquity in human genetics, (2) the use of geographic, and especially continental, classification to label populations

has increased most, but continental classification ambiguously blends racial and geographic ways of conceiving populations, and (3) no standard way of classifying populations is emerging; rather, geneticists blend population classification systems at the field, paper, and sample levels. These trends combine to increase classificatory ambiguity. We then use contextual information and interview data to make sense of these trends, showing (1) the “populationification” of human genetics has established connections between geneticists and their extra-scientific contexts; (2) continental labeling is often a way that geneticists with different types of connections can collaborate and compromise with each other; and (3) geneticists link classificatory polyvocality to their efforts to resist regulation of their research by journals and funders and to their desires to project accountability to study populations. We thus conclude that the resulting classificatory ambiguity helps geneticists bolster their scientific authority and preserve their practical room for maneuver in a context rife with potential pitfalls.

BACKGROUND AND LITERATURE

Standardizing Race in Human Genetics

A large literature traces the cultural authority of genetics (Kevles 1985; Sapp 1983; Sunder Rajan 2006), especially for its accounts of human origins and differences (Bliss 2012; Epstein 2007; Reardon 2005; TallBear 2013). Historically, genetics has had an ambivalent relationship to the race concept. After WWII, geneticists largely tried to distance themselves from ideas of race, particularly the hard-hereditarian views typical at the field’s origins (Barkan 1993; Panofsky 2014; Provine 1986; Reardon 2005). But recent research shows how commercial, medical, criminological, governmental, and social justice forces have once again driven geneticists to speak directly to matters of race (Duster 2004; Epstein 2007; Fujimura and Rajagopalan 2011; Fullwiley 2008; Nelson 2015).

Race is socially constructed and racial categories are historically variable (Omi and Winant 1994; Smedley 1999). Calling race a “floating signifier,” Hall (1997) links its enduring social power to its cultural flexibility. But this has produced practical difficulties for geneticists. Interviews and laboratory ethnographies reveal that geneticists are typically embarrassed that they have considerable difficulty articulating coherent definitions of race or racial classification, even though both are deployed in their research (Bliss 2012; Fujimura and Rajagopalan 2011; Fullwiley 2008; Lee et al. 2008).

The science studies literature has long emphasized the positive functions of standardization for the production of scientific authority and the coordination of scientific action (Bowker and Star 1999; Mackenzie et al. 2013; Timmermans and Berg 2003; Timmermans and Epstein 2010). Indeed, much research on geneticists’ recent efforts to grapple with race focuses broadly on standardization dynamics. Epstein (2007) shows how the “categorical alignment” of U.S. Census racial categories with biomedical research has helped geneticists and others connect their science to the interests of multiple stakeholders, thus bringing together scientific, political, bureaucratic, and activist forms of power. Responding to these demands, international genome projects have turned to racial classification to create the field’s mainstay DNA biobanks, thereby producing a racialization of the field (Hamilton 2008; Reardon 2005; Smart et al. 2008). Likewise, pharmaceutical companies’ efforts to target racial markets (Kahn 2012; Pollock 2012), criminal justice agencies’ eagerness to build DNA databases (Duster 2004), consumer ancestry companies’ ethno-racial heritage tests (Nelson 2015), and even researchers’ justice-driven commitments to include racial minorities in research (Bliss 2012; Epstein 2007; Fullwiley 2008) have embedded racial conceptualization, comparison, and classification in genetics research.

These trends imply that racial classification is increasing in genetics and biomedicine, perhaps leading toward a standardized way of conceptualizing populations. But

other research highlights some geneticists' ongoing discomfort with racial classification, in particular its politicized and "socially constructed" character, and their attempts to promote "genetic ancestry" or "biogeographic ancestry" (BGA) as an alternative concept (Fujimura and Rajagopalan 2011). Advocates of BGA—sometimes described by geneticists as the "objective component of race" (Gannett 2014)—seek to understand and label population samples, DNA sequences, or haplogroups as rooted in specific geographic origins, and they see this as a potential standard for the genetic description of human populations (Parra, Kittles, and Shriver 2004; Shriver and Kittles 2004; Yudell et al. 2016). Yet while BGA does transform how populations are represented, race and ethnicity slip into the ancestry concept, often barely noticed, because of long-standing habits of thought, existing technical infrastructures, and research coordination demands (Fujimura and Rajagopalan 2011). BGA is thus entangled not only with racial conceptualization, but the particularly U.S. version of racial categorization (Gannett 2014).

Biomedical research authorities have also advanced racial categorization standardization. Biomedical research supported by the U.S. Department of Health and Human Services must attempt to recruit racially diverse subjects and record recruitment according to U.S. census categories (Epstein 2007). This does not always carry over into publication, as Smart and colleagues (2008) show in their analysis of 11 leading journals' efforts to promulgate standards for the racial and ethnic labels used in articles and the disclosure of researchers' labeling practices. Audits of researchers' actual publications show rampant noncompliance with both substantive and procedural standards (Ali-Kahn et al. 2011; Caufield et al. 2009).

What does this literature on race and standardization efforts in genetics have to say about polyvocality and ambiguity in population classification in genetics? First, the overwhelming focus has been on race. Other ways geneticists think about human population

classification have been of interest mainly insofar as they are entwined with racial classification. Second, classification standardization—the alignment of genetics classifications with the racial classifications of powerful institutions—is implicated in genetics' scientific and cultural authority. Third, classificatory polyvocality is noted as a *failure* of standardization and thus a threat to the benefits standardization brings. This literature leaves no space to consider the persistence and even growth of classificatory polyvocality and the potential functions of classificatory ambiguity for geneticists.

Functions of Ambiguity in Scientific Practice

Most of the literature interprets ambiguity, polyvocality, and underlying disagreement as a problem for science. Research on scientific controversies shows that scientists are eager to settle disagreements, because individual and collective credibility are at risk as disputes persist (Collins 1985; Gieryn 1999; Rudwick 1985; Shapin 1994; Shapin and Schaffer 1985). Other studies argue that public disclosure of disagreement undermines public perceptions of scientific authority (Nelkin 1992; Zehr 2000), and there is a tight relationship between fields' scientific status and their degree of paradigmatic consensus (Whitley 1984). Finally, there is an extensive literature on the management of ambiguous or polyvocal classification through standardization—the "production of uniformities across space and time, through the generation of agreed upon rules" (Timmermans and Epstein 2010:71). By disciplining ambiguity, concealing it and constraining its spread with set categories, standardization helps quell potentially corrosive disagreement and coordinate actions of scientists and others.

A smaller literature, however, explores positive functions of ambiguity for scientific practice. Levine (1985) writes that the quest to understand and implement rationalization in modern social thought has led to serious underestimation and misrecognition of the

positive functions of ambiguity, defined as dissensus about the meaning of concepts in scientific discourse. He discusses two positive roles for ambiguity in science. The first is the “evocative representation of complex meanings . . . [that] can ignite a cluster of insights that in turn lead to novel explorations” (Levine 1985:218). Second, citing the example of “multivalent” Darwinian theory, Levine (1985:218) states that ambiguity serves the “bonding into a vital transgenerational community of a body of diverse enquirers holding somewhat different views of what are essentially contested concepts.” Ambiguity can thus grease the gears of novelty and productivity by generating ideas and solidarity.

Others have theorized the positive role of ambiguity for coordination across research communities. Centellas, Smardon, and Fifield (2014) show how interdisciplinary collaboration need not require agreement about crucial practical issues so long as there is ongoing “calibration” of practices. Star and Griesemer’s (1989) boundary objects have fundamentally ambiguous conceptualizations, because actors from distinct social worlds understand them differently. Coordinated action is made possible, not because actors reach consensus about norms and practices, but because they disregard the boundary object’s ambiguity and act as if everyone shares a definition. For these researchers, unacknowledged ambiguity helps coordinate actions of individuals from different worlds.

In a similar vein, Meloni and Testa (2014:433) argue that the vibrant growth of and scientific interest in the emergent field of epigenetics is due largely to imprecision and flexibility in its definition. They argue that, “the ability to entertain multiple understandings of what constitute epigenetic phenomena, and hence multiple ways to secure epigenetic evidence, is foundational to epigenetics’ rise, both as a discipline and as a popular phenomenon.” Their analysis extends the concept of scientific “boundary objects.” On the one hand, epigenetics is a boundary object in that different researchers understand it differently; on the other hand, it is not just an “object” but

an entire “epistemologically imprecise” domain of activity. Furthermore, where the boundary object idea implies an illusion of conceptual agreement that helps suppress or deflect corrosive controversy among different actors, the epistemic imprecision of epigenetics spurs a set of controversies that entangle researchers into activities that build the authority of epigenetics as a scientific approach.

The literature thus highlights two positive functions of ambiguity in science. First, ambiguously defined objects can enable coordination, especially among actors from different social worlds or disciplinary backgrounds. Second, ambiguity can be cognitively and epistemically generative—the unsettled character of certain scientific objects can make them intellectually productive. But these treatments of ambiguity have limitations. First, they largely depend on actors’ misrecognition of ambiguity. Part of what we will demonstrate is the explicit and acknowledged polyvocality of population classification in human genetics. Second, we will show how ambiguity can help coordinate actions not just between social worlds or disciplines, but also within a social world differentiated as a field with different clients and demands. Third, this positive literature on ambiguity largely ignores issues of power and authority. We show how geneticists use ambiguity to resist what they perceive to be excessive control by funders and journals and to establish accountability and good relations with subject populations. Our study thus helps reveal previously unrecognized positive functions for ambiguity in science.

DATA AND METHODS

To investigate the question of ambiguity in the larger molecularization of race narrative, we use a mixed methodology of content analysis and interviews. The former allows us to answer questions about *what* the patterns of population categorization have been over time, and the latter addresses *how* genomics researchers have understood the implications of these patterns and justified the practices that led to them.

Table 1. Classification Systems and Codes

Code	Classification System	Examples
1	U.S. Census (Race)	White or Caucasian, Black or African American, Hispanic or Latino, etc. (Note that the U.S. Census system combines racial and ethnic categories. We consider all of these part of the same [racialized] classification system.)
2	Continent	European, African
3	Continental region	Northern European, West African
4	Country	Netherlands (or Dutch), Japanese
5	Country region	Western United States, Sicilian, Australian state of Victoria
6	Ethnicity	Bedouin, Han
7	Language	Bantu speakers
8	Other	Usually religion: Jewish, Druze, Amish-Mennonite

Our first data source is a content analysis of the population categories used in three years of the field's top journal, *Nature Genetics*. To uncover trends in the use of population labels, we looked at all articles published in 1993 (Vols. 3–5), 2001 (Vols. 27–29), and 2009 (Vol. 41). In each of these years, we downloaded and read items presenting original research (research articles, letters, and brief correspondence, but not reviews, editorials, or news items) and recoded population labels in terms of the classificatory schemes from which they derive. We captured these labels for each of the following sections of each article: title; abstract; introduction; methods; cases, controls, and replications; tables; figures; results; and discussion and conclusion. We recorded verbatim all of the population labels used in a particular section of a particular article.

We then coded labels in terms of the larger classificatory system of which they were a part. Thus, if a paper describes a population as “black” or “Latino,” we coded this as being part of the U.S. Census racial classificatory system. If samples were described as “West African,” we labeled this as part of a continental region classificatory system. All told, we identified eight different classificatory systems in play in the articles we studied. We also sought to distinguish if a label was applied as a direct description of the population (subcode: A); a negative label, such as “not-Icelandic” (subcode: B); or “derived” from or “ancestral” to, such as “British kindred” or “European

descent” (subcode: C). Because our interest is in the classificatory system used, using negative or ancestral labels is still a way to invoke a particular system. Table 1 summarizes the classification system and codes.

These eight different ways scientists label populations come from three different logics for conceiving the essence of group belonging: racial, geographic, and ethnic. Racial classification assumes that biological peopleness is the principle of population similarity and difference. Geographic classification assumes that geographic proximity is the principle. And ethnicity assumes similarity and difference in terms of common culture (broadly conceived here to include language and religion). To track how geneticists use these different fundamental understandings, we also aggregated the detailed coding scheme into this three-part scheme.

Before explaining this aggregation, we have to discuss an ambiguity in the way geneticists refer to race in their data. As it turns out, although regulations stipulate that U.S. government-funded research collect data according to racial categories as defined by the U.S. Census (Epstein 2007), results are fairly infrequently reported in these terms (see Figure 1). Furthermore, census categories reflect both a racial logic and the parochialism of the U.S. situation (with its particular ethnoracialization of the Hispanic category, and its demographically and historically specific set of categories).

More crucially, the “continent” system for labeling populations is fundamentally more ambiguous than the other seven ways of labeling populations.¹ Continental labels blur racial and geographic understandings of population difference. When a sample is identified as “Asian,” is that a continental or racial term? “African American” is a conventional racial label in the United States, but when the label “African” is applied, is this suddenly a geographic label? Given this ambiguity, we chose to follow Gannett (2014), who argues that continental labeling has become a stand-in, or euphemism, for racial labeling of populations.² Thus, in our collapsed coding scheme we recoded the racial and continental classificatory systems (Codes 1 and 2 in Table 1) as race, other specifically geographic systems (Codes 3, 4, and 5) as geography, and the ethno-cultural systems (Codes 6, 7, and 8) as ethnicity. Despite this coding decision, we also attend to the basic ambiguity inherent in the “continental” scheme.

Our second data source is interviews, conducted at the closing period of our content analysis, which aimed to reveal the meanings, motives, and coping strategies accompanying geneticists’ labeling practices. We interviewed 36 members of the professional elite of contemporary genomics research. We selected interviewees who were widely acknowledged leaders in contemporary genomics, ran major laboratories (academic, government, or private), and published in prestigious venues including *Nature Genetics*, the subject of the content analysis. Because we solicited interviews as part of a larger project on race in genomics, we made special effort to select interviewees who had published or publicly commented on issues of human population difference and race. Genetics is a social field with complicated cleavages, hierarchies, and competitions for status recognition (Bourdieu 2004; Panofsky 2014; Sapp 1983). Our interviewees thus represent not the “average” opinion of geneticists, but the views of elites who currently dominate the terms on which “scientific capital” is most profitably earned and spent in the field. Thus,

although their views are not universal, they are particularly relevant to current patterns of authority in the field.

We conducted interviews at scientists’ localities around North America between April 2007 and June 2008. Thus, the views expressed pertain to the time when scientists researched and wrote the last set of articles in our content analysis. The digitally recorded interviews typically were between 45 and 120 minutes long. They were conducted on the record, although in this article we do not identify speakers. Interviews were semi-structured and open-ended. Questions focused on the practices and major findings of each scientist’s lab; their views of the state of the art and future directions of biomedical research; their conceptions of race and the utility of using race in biomedical research; and their views of the relationship between the knowledge they produce and their social, political, and biomedical responsibilities. Interviews were transcribed in full and coded.

FINDINGS

Content Analysis

To understand how members of the field have actually used population categories in their research, we turn to the content analysis of articles from three years of *Nature Genetics*. With these materials we can investigate the prevalence of population classification, which classifications are used most, and whether classifications in use converge toward a standard. We present three basic findings: (1) labeling the populations from which human samples derive has increased to near ubiquity over the study period; (2) the most common way to label populations over time is geographic, but the greatest growth is in “continental” labeling, which is quasi-racial and nearly indistinguishable from racial classification practically speaking; and (3) rather than standardization of labeling practices or separation of labeling logics, over time scientists have increasingly combined classifications at the field, paper, and individual sample level.

Table 2. Overview of *Nature Genetics* Sample

	1993	2001	2009
Total items	261	316	305
Research articles	189	221	204
Non-human studies	60 (.32)	88 (.40)	84 (.41)
Human studies, no population labels	67 (.35)	59 (.27)	7 (.03)
Human studies with population labels	63 (.33)	74 (.33)	113 (.55)

Note: "Total items" includes non-research items like comments, editorials, and reviews. Figures in parentheses are fractions of the research articles.

Ubiquity of assigning labels to human samples. Table 2 gives an overview of the number of articles in the sample. The number of published items in the monthly journal increased after 1993, but stabilized just above 300 in the later years, with research articles making up about two thirds of the total. Among human genetics articles, there has been a large increase in those that specifically label the populations from which samples derive. In 1993, articles about humans were roughly split between those that did and did not disclose populations. By 2009, very few human studies lacked population labels (7, or 3 percent of all research articles). Thus, the first finding is the great increase, almost to ubiquity, of population labeling in human research.³

Prevalence of geographic and continental labeling. Figure 1 looks at the frequency (among articles with human population labels) of any of the eight classification schemes anywhere they might appear in an article. Racial classification declines over the years, but we see a tremendous increase in the use of continental labels. Thus, strictly racial labeling (in the sense of U.S. census-type categories) decreased while continental designations, often euphemisms for race, surpassed them. Recall also that continental labeling is the most inherently ambiguous—it is imprecise geographically and it muddles the race/geography distinction. The biggest change here is thus a dramatic increase in ambiguous labeling.

Researchers most frequently use country labels for samples over time. The slight decline in country-based labeling in 2009

may be explained by substitution for continent-based labels. The small increase in continental-region labels may also be racial euphemisms, because labels like West African and Northern European can be synecdochic stand-ins for racial groups. Finally, ethnic, linguistic, and religious labels are rarely used over the years.

Figure 1 looks at each classificatory possibility distinct from each other, but what happens when we collapse these eight schemes into their basic definitional logics? Figure 2 shows the frequency of the use of ethnic (6, 7, and 8), geographic (3, 4, and 5), and racial (1 and 2) schemes. As discussed earlier, we followed Gannett (2014) and coded continental labeling as implicitly racial. The biggest story in these data is the dramatic increase in the use of racial classification by 2009. But this is driven largely by the increased prevalence of continental-based population labels (category 2 in Table 1), rather than use of the U.S. Census' racial labels. Thus, another way to look at this is as a dramatic increase in different kinds of geographic labeling, which are racialized or racial synonyms to differing degrees. Put differently, racial categorization is increasing, but only by making it more ambiguously muddled with continental geographic labeling.

Indiscriminate combination of classification logics. Our third finding is that geneticists mix classification systems at the level of the paper, the field, and even the sample. A good example of what this classification mixing looks like in practice is found in Keinan and colleagues (2009), which compares variations among the X chromosome

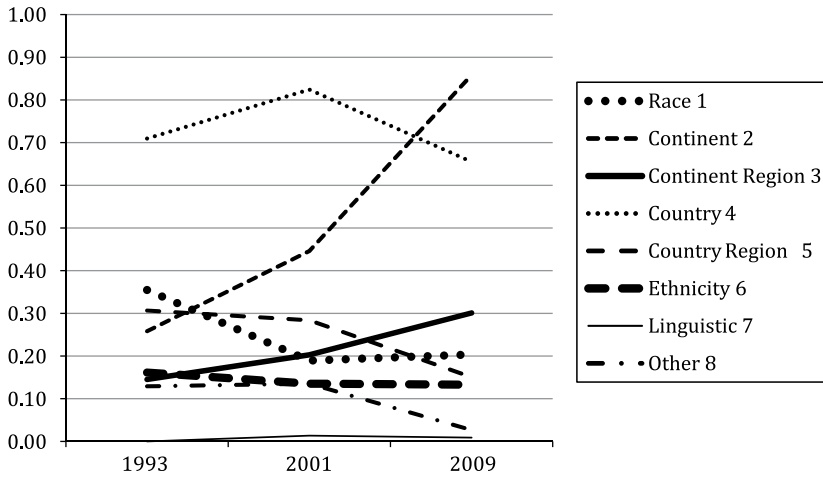


Figure 1. Frequency of Each Classification System among Articles with Population Labels

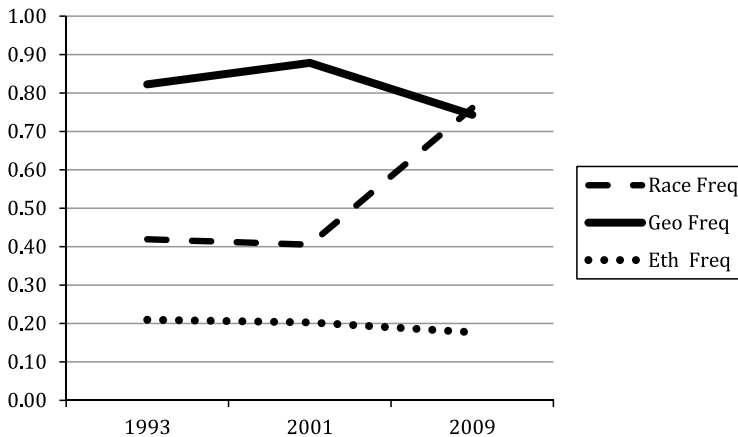


Figure 2. Frequency of the Three Collapsed Classificatory Systems

Note: Articles that use more than one type of classification are counted in each line, thus the sum of the counts in each year is greater than the total number of articles.

and autosomes to find evidence of evolutionary forces in the ancient migratory history of humans. Table 3 displays the population labels used in the various sections of the article and how we coded these labels with our full and collapsed coding systems.

This paper, more complicated than most but illustrative nonetheless, uses labels from five different classificatory systems (race, continent, continental region, country, and ethnicity) from each of the three logics for conceiving populations (racial, geographic,

and ethnic). Populations represented according to these different systems are discussed and compared directly within various sections of the paper, for example: “the autosome-to-X drift ratio comparing North Europeans and East Asians . . . and Chinese and Japanese . . . are both consistent with $\frac{3}{4}$ ” (Keinan et al. 2009:68). These labels are used irreflexively and with no discussion of the different classificatory orders they represent.

We also see the ambiguous character of particular labels and how, at times, their

Table 3. Coding Example Demonstrating Multiple Classification Logics

Section	Population Descriptions	Population Codes	Collapsed Codes
Title	“out of Africa” ^a	0	
Abstract	East Asia	3a(1a)	Race
	Europe	2a	
	Africa	2a	
	non-African	2b	
Introduction	non-African	2b	Race, Geo
	West African	3a	
	North European	3a	
	East Asian ancestry	3a(1a)	
Methods	West African	3a	Geo, Race, Eth
	North European	3a	
	East Asian	3a(1a)	
	European American	2a	
	Han Chinese	6a 4a	
	Japanese	4a	
	African Americans	1a	
	Nigerians	4a	
Tables	North European	3a	Geo, Race
	East Asian	3a(1a)	
	West African	3a	
Figures	West African	3a	Geo, Race
	North European	3a	
	East Asian	3a(1a)	
Results	West Africans	3a	Geo, Race
	non-Africans	2b	
	North European	3a	
	East Asia	3a(1a)	
	Chinese	4a	
Discussion	Japanese	4a	Geo, Race
	West Africans	3a	
	North Europeans	3a,	
	East Asian	3a(1a)	
	Japanese	4a	

^aThe title uses the phrase “out of Africa,” but because this term is not referring to a population or group, we did not give it a code.

logical register varies with the context in the article. A case in point is the label “East Asian.” In the abstract, this label is clearly being used in the racial/continental register: “around the time of the dispersal of modern humans out of Africa, chromosome X experienced . . . no similar patterns associated with the dispersals into East Asia and Europe.” The implication is the analysis pertains to major ancient population migrations that produced the continental populations that are often thought of as the three “major” racial categories. But in the methods section, “East Asian” is used in a manner more consistent

with continental region, through comparison to West African and North European populations (although racially associated, this specification acknowledges the good deal of within-race or within-continent variation among, for example, West and East African populations).

Furthermore, the methods section reveals synecdoche, whereby smaller populations defined by distinct logics are taken to be representative of larger ones. Claims about “Africans” are thus based on a “West African” sample that is actually made up of 120 people from “Ibadan, Nigeria.” “European”

claims are based on a “North European” sample composed of 120 “European American chromosomes” from individuals of “North European ancestry” from “Utah, USA.” Later, an “African American” male sample is discussed, although without information about its geographic origins. The point here is not to dwell on the kinds of historically and spatially specific evidence that geneticists use to make sweeping claims about human history and relationships. Rather, we note the profligate ways that different kinds of population classification systems are combined and related and the multiple meanings that specific labels can have depending on context.

Figures 1 and 2 show the total use of classification systems but do not say anything about how they are used together within articles. Figure 3 shows the fraction of human population-labeled articles that use labels somewhere in the text from each combination of the collapsed codes. For example, Keinan and colleagues (2009) counts as one article that used the racial, geographic, and ethnic classification systems. Across the years, researchers used all classificatory systems and all possible combinations in their articles. The popularity of the classificatory systems varies. Ethnicity is consistently the least popular, and geography is overall the most popular. The increase in the use of the racial classificatory system between 2001 and 2009 is due mostly to the increased use of race alone (again recalling our continental coding), as well as its increased use in combination with geography, and there is a minor increase in the use of all three systems.

These data have striking consequences for how we should view geneticists’ conceptualization of a population. In 1993 and 2001, just about 60 percent of articles stuck to a single classification system. Whatever system these papers were using, they were at least consistent about what a population is—it is unambiguously a racial group, a geographic group, or an ethnic group. But in 2009, just over 40 percent of articles defined populations in a single way (and even some of these, of course,

were using the ambiguous “continent” classification). In a story where geneticists are increasingly concerned with a conceptually precise understanding of what a population is, we would expect increasing purification over time. Perhaps one system would expand toward domination, or, if geneticists are deciding that these different modes of human belonging have their own logic and analytic utility, systems could become more distinct from each other. Here we have the opposite: increased *combination* of classificatory systems within articles over time. This suggests that the population concept is being deployed more ambiguously over time among geneticists and within articles.

Figure 4 considers the classification labels used among *all* the articles studying human subjects (Figure 3 omitted the articles that did not label human populations) and combines the combinations that we previously separated. Apart from the major shift toward labeling human populations, the other big shift revealed here is toward classificatory combination. In 1993 and 2001, about 20 percent of human research articles combined classification systems, but in 2009 over 50 percent did. Population labeling has become standard practice, but no standard way of doing so has emerged, and researchers have combined systems indiscriminately.

A final way to look at the combination of classification systems is through the use of hybrid labels, or labels that use elements of two different classification systems, to designate a single population sample. In the 250 articles with human population labels in our corpus, 51 used a hybrid label. The prevalence was essentially flat over time: 24 percent of articles with human population labels in 1993, 18 percent in 2001, and 20 percent in 2009. Table 4 shows all the different ways classification systems are combined into hybrid labels in our sample, with examples of the original population labels.

Some of these hybrids are relatively conventional—Han Chinese or Ashkenazi Jew, for example—in that the more specific label (Han, Ashkenazi) almost always appears with the

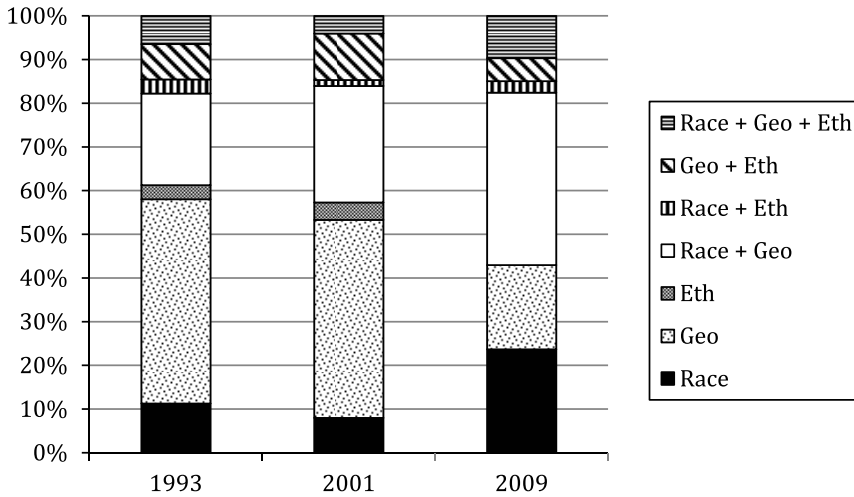


Figure 3. Percentage of Articles (with Any Population Labels) with Possible Combinations of Classificatory Systems

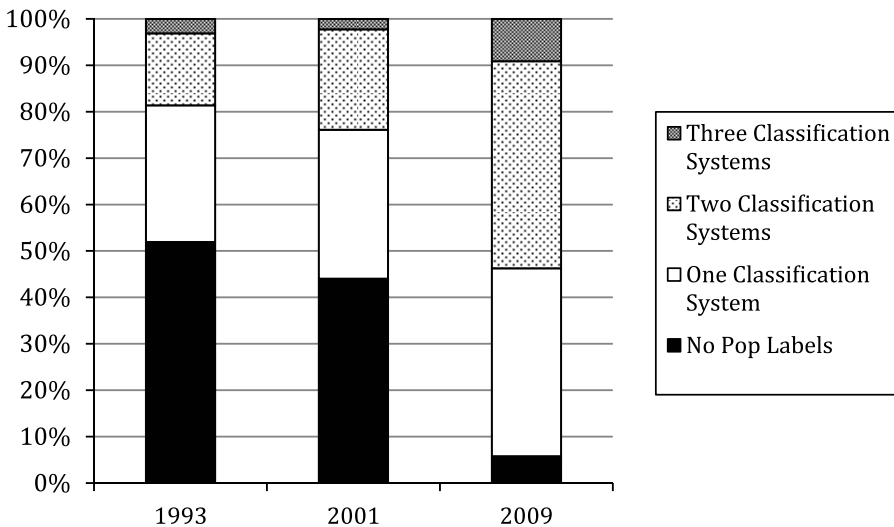


Figure 4. Percentage of All Human Subject Articles Combining Classification Labels

other. Other hybrids seem to acknowledge the diversity of multicultural countries, such as “Germans of European descent” (4a|2a) or “Utah Caucasians” (5a|1a). Others seem redundant, such as “Yoruban African” (6a|2a) or “White European ancestry” (1a|2c), but perhaps designations like the latter represent an implicit rejection by some geneticists of the idea that racial and continental labels are both racial. Most of the hybrids are driven by the

desire to specify population origins as much as possible—the most extreme example being the “Amharic- and Oromo-speaking Ethiopians” (4a|7a). Cases like these seem to be exhibiting what some interviewees called for (see the next section), that is, detail about the character and collection of the sample. Some researchers are clearly aiming for precise descriptions, but these practices are too sporadic to represent a standardization of process.

Table 4. Examples of Hybrid Labels Used, 1993, 2001, and 2009

Hybrid Label Codes	Collapsed Code	Population Description Examples
1a 2c, 2a 1a, 2a 1c	Race/Race	White European ancestry, European Caucasian, European Caucasian ancestry
1a 4a, 1a 4b, 4a 1a, 4a 1c	Race/Geo	Caucasian families from France, Asians (Chinese, non-Chinese), French Caucasian, South African family of Asian origin
1a 5a, 5a 1a	Race/Geo	Utah Hispanics, Utah Caucasians, Southwestern American Indians
2a 4a, 2a 4c, 2c 4a, 2c 4c, 4a 2a, 4a 2c	Race/Geo	Indian Asian; North American families of English or Welsh descent; African-Brazilian descent; Indian Asian ancestry; Germans of European descent; Scottish schizophrenics of European descent
2c 5a	Race/Geo	African ancestry in the Southwest United States
3a 2c	Race/Geo	North Americans of predominantly European ancestry
3a 4a, 4a 3c	Geo/Geo	north-European British; Canadian families of northern European origin
3a 8g, 3b 8g	Geo/Ethnic	Middle Eastern Jews, Middle-Eastern non-Jewish
4a 5a	Geo/Geo	Canadians from metropolitan Toronto
4a 7a	Geo/Ethnic	South African Bantu speakers, Amharic- and Oromo-speaking Ethiopians from Shewa and Wollo provinces collected in Addis Ababa
4a 8g, 8g 4a	Geo/Ethnic	Israeli-Druze origin; Old Order Amish in the United States
5a 3c	Geo/Geo	Utah residents with northern and western European ancestry
6a 2a	Ethnic/Race	Yoruban African
6a 4a	Geo/Ethnic	Arab-Israelis, Arab-Bedouin families from Israel, Ashkenazi-Israeli, Han Chinese
6a 5a	Geo/Ethnic	Chinese Han population (Central and Southern China)
6a 8g, 6c 8g	Ethnic/Ethnic	Ashkenazi-Jewish, Ashkenazi Jewish ancestry

Overall, these hybrid labels show that different logics of population labeling are combined at the sample level, not just paper and field levels. Furthermore, they highlight an ironic tradeoff between labeling precision and conceptual ambiguity. Hybrid labels may more precisely describe the origins of a population sample, but they often do this by combining logically distinct ways of conceptualizing population. Empirical specification may lead to conceptual ambiguity.

To summarize, about half of human samples had population labels in the earlier periods, but nearly all did by the end. Of the eight different ways to classify populations, country-scale geography is the most common, ethnic

labeling occurs consistently in about one fifth of articles, and continental labeling has increased in the latest period from 26 to 86 percent. We marked this as an increase in racial categorization. But more crucially, continental labeling is inherently ambiguous, mixing racial and geographic logics, and so the biggest change is an increase in the most ambiguous classification system. Furthermore, about one fifth of papers use hybrid labels that may be descriptively precise but combine classification schemes in logically ambiguous ways. Overall, combinations of classification systems have become more frequent across the field and within papers. Clarification, purification, and standardization of

classification practices are not the trend. Rather, combination, hybridization, and ambiguity in human population classification—classificatory polyvocality—have increased over time.

Interview Data

We now turn to interview and other data to contextualize and explain the three basic findings of the content analysis (labeling ubiquity, primacy of racial/continental labeling, classificatory ambiguity) with three further findings: (1) the ubiquity of population labeling has facilitated connections between geneticists and their extra-scientific contexts; (2) scientists use continental labeling to compromise and build bridges across situational boundaries; and (3) classificatory polyvocality affords geneticists the ability to resist regulation of their research by journals and funders and to promote project accountability to study populations. By reading genetics as a social field (Bourdieu 2004) whose members pursue different strategies for acquiring scientific capital, which demands balancing relationships with each other and connections to different contexts outside genetics, we can see the increase of population labeling as part of the transition from the dominance of molecular biology to the rise of human genomics, with its emphasis on population comparisons and computational methods. Racial/continental labeling and mixing of classification systems are the products of geneticists' strategies for coping with some of the pressures and dependencies of the field's situation. In short, geneticists strive to achieve a form of authority devoted to preserving collective and individual room for maneuver and professional and scientific freedom. Through such activities, geneticists valorize a version of scientific research that emphasizes short-term productivity and descriptive precision in labeling, rather than long-term consistency and standards.

Ubiquitous population labeling and the transformation of genetics. Before

turning to our interview data, we interpret the rise in population labeling in human samples as one sign of a longer-term transition within the field of genetics. In 1990, the Human Genome Project (HGP) was launched to produce a reference sequence for human and comparative genetics. The HGP catalyzed rapid technological development that made DNA sequencing and whole genome assessments of genetic variation outsourceable and relatively inexpensive. Scientifically, this meant human genetics increasingly measured DNA, but instead of molecular mechanisms of gene function, the focus was identification and comparison of DNA sequences within and among human groups. A large proportion of human genetics has thus become populational and computational, and it demands huge numbers of subjects for adequate comparisons.

This techno-intellectual transition is linked to professional reorganization. First, it requires a much larger scale and more integrated research infrastructure (in comparison to lab-based molecular genetic research). Successful research depends on data from many thousands of individuals and comparisons of far-flung populations, and these are difficult for any one group to compile. To acquire data, multi-site collaborations among groups have become necessary. Researchers are also increasingly dependent on the goodwill of study populations, and they utilize collective resources like biobanks and data repositories. Furthermore, the population push in human genetics has linked researchers to different domains of application, in particular, medicine, pharmaceutical development, forensics, and nascent direct-to-consumer genetics.

The "populationification" of genetics has also been spurred and framed by a set of political and bureaucratic forces. A long-developing movement for a more inclusive biomedical research establishment achieved a major success in 1993 when President Clinton signed the NIH Revitalization Act, which set into motion a series of reforms spurring more inclusive research and drug development. Researchers were required to use OMB

census categories in their subject recruitment and data collection. These changes spurred debate among scientists and science policy-makers about population classification practices; this debate was likely a causal factor in the overall rise of population labeling we see in our quantitative data.

Journal editors have sought to standardize population classifications, and especially transparency about labeling practices (Smart et al. 2008). Major journals revised their “uniform requirements” standards for manuscript submission in 1997, calling for care in the use of race and ethnicity because these categories “are ambiguous” (Ali-Kahn et al. 2011; Epstein 2007). Some went so far as to call race “unscientific” and to inform researchers not to use these categories without a specific scientific rationale.

In 2000, an editorial appeared in *Nature Genetics*, the field’s leading specialist journal, calling on scientists to “explain why they make use of particular ethnic groups or populations” and to consider whether their work was reifying race as a genetic concept (Anonymous 2000). The editorial not only asked researchers to clarify and rationalize their population labeling procedures, it suggested that “race” was pseudo-biological and the field would be better off abandoning racial labeling for more precise labels.

Journals were not of a mind about what to do about racial classification. In 2003, Bette Phimister, a *New England Journal of Medicine (NEJM)* deputy editor (and *Nature Genetics* editor until 2002), published a directive tasking researchers to use racial standards: “it seems unwise to abandon the practice of recording race when we have barely begun to understand the architecture of the human genome and its implications for new strategies for the identification of gene variants that protect against, or confer susceptibility to, common diseases and modify the effects of drugs” (Phimister 2003). Following the spirit of the inclusion movement, *NEJM* made the opposite call: racial categories should be used and tracked to better understand health and treatment disparities.

Nature Genetics changed course somewhat in June 2004, asking researchers to shift from racial categorization to the measurement of ancestry and ethnicity. An editorial, “The Unexamined ‘Caucasian,’” defined ancestry as “descent, continental origins, and admixture” and ethnicity as the social and cultural factors that shape “phenotype, migration, and reproductive patterns” (Anonymous 2004). This move, the editors argued, would help describe health inequalities better as a result of racial discrimination. Again, they asked for stronger disclosure of the labeling procedures researchers use to facilitate the generalizability and transferability of findings. Thereafter, the journal asked authors to report “ancestry” in biogeographic terms.

Soon thereafter, the *Journal of the American Medical Association* issued a set of “author instructions” requiring greater clarity and justification in the way papers used racial categorization (Winker 2004). The editorial demanded transparency on these matters, while acknowledging that use of population labels is pulled by many competing demands, such as research questions, changing demographics, community preferences of research subjects, and funder mandates.

We have shown how the “populationification” of human genetics is linked to a set of scientific and professional conditions to which researchers had to adapt in the period circa 2009. In contrast to strictly molecular research, the new style of research demanded multi-group collaborations and collective data resources; it strengthened researchers’ ties to various contexts of application but also their dependence on study populations to participate in research; and it led to increased oversight efforts by funders and journals. We now turn to interviews that reveal how geneticists’ negotiation of these demands pushed the rise of ambiguity through continental/racial classification and mixing of categories.

Negotiating collaborations and clients’ demands with geographic/continental classification. What explains the rise of geographic, and in particular,

continental labeling, that is, the ambiguous form of racialization that mixes indistinguishably racial and geographic ways of describing populations, in the last period of the content analysis? Our interviews suggest that the prominence of this form of labeling is linked to geneticists' efforts to balance demands placed on their research by the institutional interests of external patrons and clients, as well as the necessity of multi-lab collaborations. Geneticists interviewed often expressed skepticism about the use of strictly racial labeling in research, yet they face many demands to speak about racial differences. For many, continental labeling is a kind of compromise: an implicit way of using quasi-racial labeling without speaking in a way that explicitly geneticizes race.

Interviewees often expressed skepticism about the relevance of racial classification to genetics research. A director of one of the largest global genome projects did so in his denunciation of the federal guidelines for classifying samples by race:

I think they are ludicrous, personally! What does "Caucasian" mean? It's everything from somebody living in Belfast to somebody living in Southern Spain to somebody living in Tunisia to somebody living in Sri Lanka. That's a *huge* range of variation. What does "black" mean or "African American"? There's more variation in the average African village than there is in the rest of the world outside of it combined.

Even a head official in public health, who has led several international genome projects, characterized federal racial labeling standards as "based in a way that is pretty hard to defend." He said the federal classification "tends to reify the concepts that those groups are biologically different, and clearly that's not defensible."

Many interviewees were supportive of the idea that geneticists should work to include racial and ethnic minorities in research, and that closing health disparities is an important research aim, but only one, a leader of minority-focused genome projects, made supportive

comments about the idea that geneticists should use racial labeling as a standard classification system. He justified standard racial labeling as sound science: "It can't be the Wild West. I mean, this is just good science. It just makes good science sense. . . . You can imagine trying to buy something, and you say, 'I want a kilo of this.' If everyone is using a different measure of a kilo, it gets crazy!" As he has done in the scientific literature, this speaker privately supports racial standardization as important to securing minority inclusion in research.

In front of this already contentious backdrop, contemporary genetics demands collaborations among research groups with conflicting norms about how to label samples and classify populations. As a chief developer of human variation technology at the top privately funded lab in the United States explained:

It's a *huge* collaboration with all these internal rules and paper-writing rules. They call African Americans "black" there. In those papers, I had these fights associated with that. I've tried to call African Americans "African Americans" the whole time, and they want to call them "black" because that's their rulebook.

Here the disagreement is less about the classification system—both black and African American are racial labels in the U.S. context—but what labels to use.

Negotiations about what labels or classifications to use are often described as contentious. The increasing use of geographic classification was commonly described as an agreeable way to settle these disputes. For example, an NIH scientist described negotiations about the term "Caucasian":

A colleague over at Hopkins . . . had used the term "European" . . . and then elsewhere in the paper she had used the term "Caucasian." And, I wrote to her as I reviewed the paper, and I said, "Look, you gotta pick one here" . . . I said, "My preference, and what I have settled on, is 'European American' and 'African American.' It's *descriptive*." Years

ago, [senior genomics researcher] yelled at me for using the term “Caucasian,” and he said, “Those are from the Caucasus! That’s over in Russia!” And, I just said, “Fine, I’ll stop using that.” And by and large, that’s worked.

This quote illustrates several key dynamics. First, the speaker describes how the conventional racial label “Caucasian” makes no sense for most samples if it is taken as a literal geographic description. Second, continental labeling has the benefits of providing a consistent system that can describe multiple samples and being “descriptive,” referencing something about the physical origins of the samples. Third, continental labeling allows the simultaneous invoking of a U.S. racial label, African American, as equivalent to a geographic label, European American. Implicitly, both labels are racial and geographic; although offering “consistency” here, continental labeling is also ambiguous between these two forms of classification. And fourth, the speaker implies that continental labeling was an agreeable settlement of the dispute.

While a settlement like this suggests the appeal of continental labeling, it is not a universal solution for geneticists. For one, geneticists’ work intersects extra-scientific domains with different demands for classifying populations. A former president of the American Society of Human Genetics (ASHG) explained:

Now in the forensics world, police and so forth, they use it [Caucasian] all the time. And the *New England Journal of Medicine* prefers “white” and “black.” So, they use the term “whites,” and some people would have trouble with that. So, there is *no* descriptor that works perfectly all the time for *anyone*.

As researchers shuttle between forensics, medicine, and academic population genetics, they must adapt to different domains that have their own preferred ways of classifying populations. Furthermore, continental labels can be too general when particular domains of application demand particular levels of precision. As the

same forensics expert explained, “Also, you don’t want to allow for inappropriate generalizations of your results, which is what happens when you say ‘African’ or when [my pharmacogenomics collaborator] says ‘sub-Saharan African.’ I have talked with him about this many, many, many times!” Here the dispute was about what kind of synechdotic inferences could be justified, and the appropriateness of continental labeling or, indeed, the quasi-racial geography of sub-Saharan or black Africa. Not only labels but also the legitimacy of classification systems for particular inferential purposes had to be negotiated.

These quotes help us understand the prevalence of geographic labeling and the growth of continental labeling revealed in the content analysis. Geneticists face a complex set of pressures regarding population labeling. Funders and journals have tried to impose controversial rules about the classification and description of samples, and the different domains of application have their own demands. Furthermore, geneticists need to collaborate across labs with different contextual demands, local practices, and epistemological preferences. What labels and classification systems to use are subject to constant negotiation. In this complex situation, geographic and especially continental labeling is often the “least bad” way to achieve most ends. Geographic labels have the appearance of neutrality among scientists’ competing preferences, and continental labels, in particular, allow scientists to actively trade on their ambiguity: geneticists can evoke some of the meaning of racial labels (demanded in many contexts) without committing to all of the problematic aspects of racial labeling. But it is also clear that many believe continental labeling to be inappropriate sometimes, so its availability as a standard is problematic.

Causes of mixing classification systems. What explains our last finding in the content analysis, that geneticists mix different logical ways of defining populations? Part of the answer is related to the points above. Some collaborations involve rationalized synthesis of labels, but in others, different

labeling schemes are simply combined in the analysis and subsequent papers. Many projects also draw from scientific repositories where data or biological samples are preclassified. Researchers must decide whether to change or directly incorporate the preexisting database labels with whatever labels the project is using.

But geneticists have several other motivations that affect classification mixing. First, with few exceptions, leading genomics researchers view journals' and funders' efforts to create substantive or procedural standards for population classifications as bureaucratic constraints on researchers' autonomy that should be resisted. Second, researchers are sharply aware of their dependence on the goodwill of study populations. One way researchers display accountability to those populations is by refusing to re-label samples. Thus, as researchers resist bureaucratic classification standards and accept people's own labels for their samples, the labels and classifications in use proliferate.

Earlier we described how many geneticists view U.S. federal guidelines governing data collection as demanding a scientifically dubious racial classification. For many interviewees, the broader problem is bureaucratic interference, especially by funders and journals. One chief director of oncological genomics at the National Institutes of Health voiced a common complaint that such standards issue from "commissions" and "committees" that "just sit around and make all these pronouncements."

Geneticists have widely divergent understandings of what the guidelines about labeling and classification actually are, but they nearly universally resent "bureaucrats" imposing rules on them. As one developer of a technology using "ancestry informative markers" (AIMs) to map admixture said:

If you can convince me that one of those labels is offensive or inappropriate, or is scientifically incorrect, then that is one thing. I suppose if you tell me I have to use the new labels or I will lose my job that

would grab my attention too. But I'm not going to voluntarily just change the labels.

This scientist expressed the prevailing sentiment that only labels derived from "what we can measure genetically" could be used across the field. But this would come from the community working in solidarity, "because there is a scientific discussion behind it, one that you can be involved in . . . so that is my answer to the labels from the bureaucrats." In fact, for this scientist and many others, anything short of an internal decision is unacceptable.

Some of the labeling policies discussed earlier were specifically about procedural standards for accurate disclosure and description of labeling practices, rather than substantive standards about what classifications to use or avoid. But interviewees often neglected this fact and conceived of standards as substantive. One leader in personal genomics, who serves as CSO for one of the world's biggest biotech firms, stated:

I think, better than just [bureaucratic] standardization, we have to explain exactly what they mean by any label. Do they mean people self-identified? Do they give people an opportunity to give multiple labels or not? Even do they give check boxes or do they have people fill in blanks? You know those—there is not any absolutely ideal world. Whatever label you use, you say how you came up about using that label.

This interviewee described frustration when reading papers whose study authors use continental terms without further explanation. She cited her then recent article with two leading bioethicists, which encouraged geneticists to define terms with as much detail as possible. Notably, this geneticist asserts scientists' autonomy by contrasting her views with bureaucratically imposed standardization, even though her views are very close to some of the "bureaucratic" labeling policies in play.

A leading African American health expert with a highly successful personal genomics company interprets bureaucratic labeling

standards as pointing toward the abolition of racial labeling of samples:

I don't think that we have to get together and have a meeting about the right label. In the end, I think if you allow geneticists to do their research, they will show through their research that [the racial label] is not clear. If you stop them, they will not. I don't even think that in this country there's a way of stopping people from using those labels. I guess you could prevent further gathering of data using the racial categories, but people will continue to use them as long as they can.

This scientist's argument is that racial labels are biologically inappropriate, but policies seeking to impose that fact upon geneticists will fail. Only free research will lead geneticists to abandon racial categorization. Note that this scientist interprets labeling policies as banning race, the opposite view of the interviewee quoted earlier who decried the bureaucratic imposition of race, but agrees that the real problem is imposing upon geneticists' autonomy.

The ambiguous mixing of different ways of classifying populations is thus partly due to geneticists' skepticism about and resistance to bureaucratic efforts to standardize population classification. Although geneticists interpret these policies in different ways and sometimes agree with their substance, they all see such policies as external and largely illegitimate. Geneticists' *laissez-faire* views on labeling are partly a defense of their authority over their own science. Through geneticists' noncompliance, these policies have largely withered on the vine (Ali-Khan et al. 2011; Caulfield et al. 2009). NIH still requires the collection of data in terms of census categories, but efforts are often *pro forma* (Epstein 2007). The research field has more successfully, although mostly passively, resisted the journal policies. Journals have not sought to enforce or update them.⁴

Mixing population classifications is more than a means for resisting external authority, it is also a means for cultivating authority in

the form of political goodwill among the populations geneticists study. Scholars have noted a "participatory turn" in contemporary biomedical and genetics research, whereby scientists find themselves increasingly accountable to the populations they study (Joss 1999; Kelty and Panofsky 2014). Geneticists have long faced charges of naïveté and arrogance for using politically contentious labels to describe populations, and for purporting to tell people who they "really are" or where they "really come from" (Reardon 2005; TallBear 2013). And the profession has gone to great lengths to cultivate a more respectful and empathetic attitude toward research participants. For example, a former president of the ASHG stated that he avoids terms that "incite negative feelings." He and others recounted the problematic history of terms like "Caucasian" and "gypsy," taking issue with social labels connoting population superiority or inferiority. Another transnational genome project director said:

I know [the label we choose] drives the way people feel about themselves. It is important in terms of the way people receive information, and it is important in terms of the way people feel about wanting to participate in studies.

Researchers see their responsibility as one that protects both subjects' rights and the field's reputation and viability with the public. They see labeling as something that goes beyond individual studies to the well-being of the science overall.

The rise in ambiguity of population labels, and the disinclination to pursue a strongly standardized way of classifying populations, becomes an ethical resource in scientists' relationships with research participants. They understand effective relationships to involve accountability to research participants' self-categorizations. Some interviewees explained that the sample population's self-reported identity in the methods section of articles is a good way to "give voice" to their subjects, and thus they may avoid generating

alternative “objective” terms. On reconciling reports with subjects’ viewpoints, the global genome project leader quoted earlier insisted:

We don’t tend to re-label people. . . . [But] in some cases, you will sample people who report to be un-admixed and turn out [to] be quite mixed with surrounding groups. And that will force us to reconsider their population history. . . . But we are *not* going to change somebody’s self-identified ethnicity.

As he explains, genetics research can often contradict people’s own sense of their identity, ancestry, origins, and migration. Researchers must therefore be careful how they describe their results. The aforementioned biotech CSO expressed the common belief that geneticists should adapt to their subjects’ political context and try to think in their shoes:

What I do is I imagine that somebody with that, who might get that label either for themselves or someone else, is reading it and can understand it. Are they comfortable with it?

When asked what would constitute the “best descriptor” for the populations they study, genomics researchers do not try to offer a strictly data-driven definition. Instead, they all replied that the social and political interpretation of labels is important to synthesize with research efforts. Some promoted geographic labels as a promising alternative. This brings us back to our second major finding. Terms like “sub-Saharan African” and “Northern European” appear specific yet flexible, they remind observers that populations are shaped by the specific environments in which they live, and they seem less politically fraught than ethnic or racial labels. Researchers are aware that geographic labels often coincide with contested cultural labels and some—“sub-Saharan African” or “East Asian,” for example—can be racial euphemisms as well, but they prefer these as seemingly neutral. The trend toward geographic labeling and quasi-racial continental labeling

we documented in the content analysis is linked to these questions of public accountability. Geography gives the impression of being objective and apolitical and thus can be seen as a way to bypass conflict with research populations.

Geography, though, has limits in many circumstances. For example, the HapMap Project distinguishes between Ibo and Yoruba, because they are communities that cohabit in Ibadan, Nigeria, yet form separate social groups. For scientific and political reasons, it is important in particular circumstances for researchers to avail themselves of multiple classification systems, so geographic standardization is not an answer to the field’s dilemmas. Flexible ambiguity of classification systems and practices, even if chaotic, helps researchers strengthen their authority by securing the consent of and avoiding conflict with study populations.

Labeling specificity as a scientific virtue. There is one more reason for these findings, especially the classificatory ambiguity. A new definition of scientific practice emphasizing descriptive precision of populations, rather than generalizable comparison, has accompanied the changes discussed here. Geneticists’ resistance of bureaucratic standards and accountability to subject populations help secure their scientific authority, but these factors also drive ambiguous mixing of classifications. Researchers have also come to see classificatory flexibility as a scientific virtue. Standard categories would not fit all the circumstances of different scientific projects, and they might make interactions among research communities with different labeling practices difficult. Geneticists thus emphasize precision over consistency in labeling.

Echoing the precision emphasized in many of the quotes used here, one former president of the ASHG concisely voiced a common sentiment, “I think we are always trying to sharpen our terminology, refine it, and make it as accurate and descriptive as possible.” As a leader of one of the most-cited studies on population variation put it:

When you are trying to investigate some particular aspect of human variation it depends on either geographic variation, or ancestors of groups, or socially defined race. What is important is to identify what is the concept of human variation that is relevant to that particular situation, and to as much as possible, to measure that specific concept.

Thus, rather than commit to a standard way of conceptualizing populations, interviewees said that researchers should create sample taxonomies based on each study's unique research questions, obtain as many layers of information about subjects as possible so that many labels can be circulated about the same data, and report with classifications that are beneficial to the groups under study. A leader in public health genomics at the NIH, for example, said that a detailed description would be superior to standard categories: "Just describe what you think you have, and what you will use it for in your study."

A strong rationale for flexible, combinatory classification can be seen in this public health genomics expert's description of evolving labeling practices as his project and the questions asked develop:

At the beginning stages, I try to make sure that everybody knows where these people are really coming from. And then later on, I can say West Africans. . . . But at some point, when we want to describe a diaspora, for example, which is the recent migrational patterns, then the concept of "Africans" and the "African diaspora"—sometimes the word "black" and just given the way we have defined that—captures that.

Evoking at one stage a very local label, later a broad geographic one, and finally a racial label is, for this expert, a function not of changing samples but changing scientific arguments and exposure of the project.

To summarize, an important way geneticists justify their use of combinations of geographic and quasi-racial classifications, and the inconsistency and ambiguity in their

labeling practices, concerns scientific values of precision and evolving research questions. Conflicting ideas about labeling present challenges to collaboration, but interviewees overwhelmingly see the solution as case-by-case negotiations among collaborators. Our interviewees expressed almost no desire to institutionalize collective standards, nor did they express consistency as any kind of ideal in labeling. Geneticists see the problem as one of pragmatic coordination, not abstract conceptualization. That is, they ask, "What are the right labels for a particular set of samples combined for this research question given the proclivities of these researchers?" not, "What is the correct conceptualization of population?"

DISCUSSION

This article aimed, first, to characterize geneticists' human population labeling practices, and, second, to analyze how geneticists describe and justify these practices. Our content analysis of human population labels used in *Nature Genetics* articles in 1993, 2001, and 2009 found (1) labeling the population origins of human samples has become almost ubiquitous over the period, (2) geographic labeling has been most prevalent and continental labeling has increased the most, but (3) these systems, despite their growth and prevalence, are *not* emerging as standards, rather, geneticists mix and combine different classification systems and geographic, racial, and ethnic logics of classification at the field, article, and sample levels. We used interviews and contextual information to situate these findings and conclude that (1) "populationification" is linked to changes to the genetics profession: scaling up of research and coordination among groups, increasing ties to clients, and political demands for diversity and inclusion of research subjects and topics; (2) geneticists explain the prevalence of geographic and continental labeling in terms of the need to balance the coordination of research groups with different labeling norms and the demands of clients who often expect racialized knowledge; and (3) geneticists perceive the mixing of classification logics, in

part, as reasonable resistance to bureaucrats' efforts to regulate research and as part of their good faith efforts to remain accountable to study populations who may object to being relabeled. While acknowledging that the ambiguous picture of population emerging from genetics might look problematic, researchers have advanced a definition of good science that favors descriptive specificity of populations over consistency and conceptual coherence.

Molecularization of Race

Both science studies observers of human population research in genetics and critical scientists within the field have warned about the racialization of genetics and the molecularization of race. An implication of this literature is the growth of racial classification in genetics papers. At first glance, our research provides support for this idea (Figures 2 and 3). Many journals (including *Nature Genetics*) have asked authors to use racial classifications less frequently and with more reflection and justification. Instead, geneticists have used ambiguous continental, geographic terms without explaining their decision within papers. This has the effect of implicitly expanding the use of race. Rather than strict racialization, however, we think this trend is better understood as an increase in ambiguous labeling. Continental labels, after all, are an unclear mix of the geographic and the racial (Gannett 2014). And their use, as our interviews show, is linked, at least in part, to confusion and compromise among geneticists about what can be agreed upon by different research groups, what is unlikely to upset research participants and activists, and what labels are appropriate to the science.

In human genetics *how to define populations* has become a fundamentally ambiguous matter. This implication, taking the perspective of the field's labeling practices and research corpus, resituates the racialization/molecularization thesis rather than contradicting it. Racialization is happening within a swirling mix of classifications. Even as the peculiar continental/racial scheme is ascending,

it is being combined with other classification systems rather than eliminating them. Different geographic and ethnic classifications are in play within the field and within papers. Hybrid labels that combine different classificatory logics in a single population are also frequently used. The indiscriminate mixing of logically distinct classificatory systems occurs at the field level, the paper level, and the sample level. Geneticists have implicitly decided that comparing populations characterized by different logics of belonging (racial group, geographic location, cultural commonality) is a routine and meaningful activity.

The literature demonstrates how the molecularization of race and the racialization of genetics is driven by political commitments, technological affordances, and client demands for racial knowledge. But ambiguity of population classification is driven largely by a set of interactions among geneticists as they adapt to their field's dependencies on a set of internal and external conditions. Rather than collectively working to establish a standard way of classifying populations or a rubric for the application of different systems, geneticists negotiate arrangements locally. Geneticists are frequently concerned about the meaning and coherence of the classification schemes and population labels they use, the decision of what labels to use for particular populations, the difficulty of assigning individual subjects to labeled groups, and the meaning of comparisons among groups thus labeled. But ambiguity persists, in part, due to the positive functions classificatory polyvocality plays—enabling collaborations, satisfying clients, resisting regulation, and respecting participants. It is further promoted by the emergence of descriptive precision as a scientific value, as the addition of more kinds of labels makes just what a population or a comparison *is* more ambiguous.

Ambiguity Bolstering Authority

Ambiguity has received a limited treatment in sociological theories of scientific knowledge production, conceived narrowly in terms of social worlds bridging and epistemic

openness. The results of this study expand sociological theories of ambiguity to include how it functions to support scientific authority. Labeling choices and self-justifications that increase ambiguity of population classification allow researchers to quell conflict and coordinate actions within the social world of genetics, while satisfying client demands, resisting regulations, and respecting research participants. All of these functions help bolster geneticists' scientific authority generally and their cultural authority over knowledge of human populations and differences.

Many factors have driven geneticists' growing scientific authority over definitions of human population, but the contribution of ambiguity has not been noted previously. Although geneticists today acknowledge that they live in a pre-interpreted world, for much of the middle twentieth century they debated ways of "objectively" characterizing human populations. Only after activists advocating for studied populations resisted important projects have geneticists become widely responsive to their demands. Furthermore, ambiguity may help geneticists avoid conflicts with academic critics and amongst themselves. The long history of intellectual struggles to define race suggests that every classification system aspiring to universality (i.e., to be *the* standard) is open to manifold criticisms, but currently the field uses many rather than standing behind one. Dwelling in ambiguity about how to classify and compare populations allows geneticists to do their work without presenting a hard target for criticism. Ambiguous labeling draws criticism to geneticists' practices and ways of portraying populations, but it also presents a critical target that is slippery and complicated to engage. Levine (1985) wrote that insofar as ambiguity is about the concealing of one's true intentions, it has no place in science. But here the ambiguous use of classification systems substitutes for a positive concept of human population, for the time being at least, and thus it serves geneticists' authority as a functional consequence but not an intentional deception.

Much of the sociology of scientific knowledge tends to see ambiguity as corrosive of scientific authority and an embarrassment to be concealed. Shapin (1994) understood an essential function of the laboratory and forms of scientific testimony to be the protection of the ambiguity of experimental practice from corrosive scrutiny. Collins (1985) understood the authority of experiment and replication to be dependent on most scientists' temporal and social distance from the ambiguity in which "core set" researchers dwell. In this tradition, ambiguity is a liability to be managed, but in our study, ambiguity is an open issue used both unwittingly and strategically in ways that contribute to collective authority. Our finding that classificatory ambiguity can be a cultural, social, and scientific resource for building authority—beyond whatever embarrassment it may evoke—is counterintuitive and should stimulate exploration of similar situations in other domains.

Our study also cautions against some of the more positive treatments of ambiguity in the literature. Star and Griesemer (1989) show how the ambiguity of boundary objects can facilitate collaborations or connections of scientists and actors from different social worlds. Levine's (1985:218) "evocative representation of complex meanings" and Meloni and Testa's (2014) "epistemology of the imprecise" turn on the idea that ambiguous concepts can energize scientific practice. The story of ambiguity due to classificatory polyvocality in genetics is more ambivalent in this regard.

In accordance with these ideas, ambiguity allows geneticists with different data sources and ways of labeling and conceptualizing populations to work together productively. When they are forced to confront differences, they settle the matter locally and temporarily rather than engaging a broader meta-methodological debate about standardization or a conceptual debate about the ontology of "groups" in population genetics. Levine (1985:17) wrote that "toleration for ambiguity can be productive if it is taken not as a warrant for sloppy thinking but as an invitation to deal

responsibly with issues of great complexity.” Ambiguity greases the gears of scientific activity in human genetics, but *pace* Levine’s optimism, does not necessarily force serious engagement of complexity. The periodic reminders of geneticists’ duties to accurately disclose labeling procedures, discuss the implications of labeling decisions, and abandon imprecise labels (such as race) might be signs of engagement of complexity (Shields et al. 2005; Yudell et al. 2016). And the fact that social scientists, public health researchers, and others have joined critical geneticists in making these calls might be evidence that ambiguity has opened up channels between social worlds or communities with different epistemological toolkits. But these calls—with their repetition and frustrated tone—along with some geneticists’ self-justifications, seem to us to indicate geneticists’ intransigence, or at least the mainstream’s practical indifference to engaging the conceptual issues of population ambiguity, and their preference for getting on with the research. The engagement across intellectual social worlds, although perhaps productive for the participants, seems confined to the margins.

Rather than a form of ambiguity that bridges social worlds or fuels dynamic intellectual hybridization, population labeling is acting more like a “boundary expressing symbol of community,” in the sense discussed by Cohen (1985). For Cohen (1985:21), “symbols are effective because they are imprecise.” Thus, geneticists all agree that “population” is among their core concepts and it must be treated carefully, but in fact there is great dissensus about exactly how population should be defined and treated in practice. “Population” is like a symbol in Cohen’s (1985:21) sense in that it aggregates perspectives and “continually transforms the reality of difference into the appearance of similarity. . . . It unites them in their opposition both to each other, and to those ‘outside’ . . . It thereby constitutes, and gives reality to, the community’s boundaries.” This helps us see how geneticists can disagree without feeling it necessary to engage or resolve the

disagreement, and how they can implicitly share a community of practice and more or less ignore those (i.e., critics) outside that practical community.

Given our association of science with rationalization and disenchantment, we normally think of it having little place for ambiguity. Ambiguity is seemingly opposed to good scientific knowledge, and we think of it as a source of deficiency and weakness in science. Our study demonstrates the opposite in human genetics. Rather than something they have sought to contain and minimize, ambiguity manifests in many different ways and has expanded in research on human populations. Rationalization or standardization has not been the route to authority in this case, but rather dimensions of ambiguity in population classification have helped human geneticists build authority through coordinated activity, resisting oversight, and maintaining accountability to clients and participants. It has been a near article of faith for the sociology of scientific knowledge that controversy, dissensus, disagreement, lack of coordination, lack of standards, and the like undermine scientific authority or power. Either they corrode credibility or represent breaks in networks of associations. But our research suggests that the non-contentious dissensus about how to represent human population functions as a source of contemporary geneticists’ authority and increasing jurisdiction: ambiguity stands for dissensus and disagreement that produce authority by providing symbolic resources for dispelling contentious controversy.

Implications for Genomic Science and Governance

Critics of labeling practices in genomics often frame their calls for standard labels, greater transparency, and poor disclosure practices in terms of scientific quality (Ali-Khan et al. 2011; Caulfield et al. 2009; Yudell et al. 2016). Indeed, the ambiguity we document should undermine the reliability and generalizability of findings in genome science.⁵ The basic policy implication is that translational

claims of genomics should be tailored to specific populations under study, not some putative general population.

Two major scandals of contemporary genomics research have been the spotty replication record of claims and the “missing heritability” problem, wherein genomic studies have been able to identify DNA associations that account for only a fraction of the variance that twin and family studies predict is due to genetic causes (Manolio et al. 2009). Genetic explanations for racial disparities in disease have fared particularly poorly in reliability and robustness tests (Ioannidis, Ntzani, and Trikalinos 2004). These problems may be partly due to population labeling ambiguities that are hindering apples to apples comparisons across papers.

An example demonstrates the value of *disambiguating difference* in genetic research. In biomedical research, “race” is often deployed but rarely rigorously specified, and thus it becomes an implicit proxy for a variety of possible genetic, social, behavioral, cultural, and political causes (Gravlee 2009). In a study of hypertension in Puerto Rico, Gravlee, Non, and Mulligan (2009) used a model that compared two definitions of race: socially ascribed color or African genetic ancestry markers. When “race” was measured only via genetic ancestry, hypertension inequality appeared to have racial/genetic causation (i.e., “African genes” caused hypertension). But the genetic ancestry association with hypertension became insignificant when a significant color/SES interaction was included. Furthermore, only in the full model where genetic and social differences were disambiguated could the effect of a particular candidate gene for hypertension appear. Here is an example where engaging the complexity of human difference, although not reducing or standardizing it, greatly improved the identification strategy. However, the dominant strategies to address the scandals of genomics have been brute force increases in sample size and deploying more advanced technologies that enable finer-grained genomic analysis (Panofsky 2015).

Although ambiguous population labeling may hinder generalizability, it may aid the portability of results into domains of application. Paired with racially stratified global biobanks, “admixture-mapping” technologies that engage in genetic “population stratification,” especially by continent, are proliferating racialized genetic correlations to a wide variety of health and phenotypic outcomes (Bolnick et al. 2007; Fullwiley 2008). These practices and technologies have spurred (and in turn been encouraged by) a range of race-based biotech investments, including drugs (e.g., BiDil and Iressa) purported to be race specific, “DNA PhotoFit” software that claims to build a digital “mugshot” of African descendants from genetic material, and DNA Talent Tests aimed at white and Asian parents (Bliss forthcoming; Kahn 2012; Ossorio 2007). These products are based on statistical genetic correlations, which do not take into account the full range of possible biological or social processes (Ossorio and Duster 2005). The lack of standards for disambiguating types, and thus causes, of population difference has led genetics research to be carried along with the racialized expectations of forensic and pharmaceutical biotech development, and disciplinary anomie around these issues may have discouraged oversight and critique from within genetics.

This study also helps illuminate the governance problems around race and population designation in genomics. The literature is littered with calls for clearer labeling practices and more care with the race concept, as well as editorial statements and empirical studies charging rampant noncompliance with these recommendations (Ali-Khan et al. 2011; Caulfield et al. 2009). The latest has asked for the National Academies of Science to convene experts to devise standards for biological researchers to “move past the use of race” (Yudell et al. 2016:565). These calls assume that the lack of effective standards in labels and labeling practices is incidental to research in human genetics. Our study shows that ambiguity is not incidental, but rather is intimately related to the way geneticists’ authority has been constructed and maintained.

Thus, public pronouncements in this domain will likely continue to be perceived as unwelcome impositions, and ambiguous labeling will continue to be the practical response. This strategy is unlikely to produce effective reform. Instead, reform would seem more likely to come from geneticists able to attach the complaints to new ways of doing research within the legitimately recognized terms for valorizing scientific capital within the field. In other words, geneticists valorizing a new research agenda that connects *disambiguation* (if not standardization) to more effective science epistemically or practically (e.g., medical or commercial success) would seem to be the most likely opening for a meaningful move past conceptual ambiguity about population classification.

Conclusion

By combining a novel content analysis with interview data, we added to the literature on the molecularization of race in genomics research and theories of scientific authority. Geneticists increasingly characterize human populations with the ambiguous continental/racial classification system. But they also proliferate and combine logically distinct classification systems to describe and compare human populations. This rise in the ambiguity of human population classification has helped the field of genomics secure its scientific authority and build its community.

Future research should consider additional contexts for population labeling practices in genomics research. Are the patterns the same in other journals within and beyond genetics? Do labeling practices differ in terms of the purposes of research (e.g., medical vs. non-medical) or the composition of research teams (e.g., U.S., international, multinational)? How do processes of synecdoche occur—that is, what smaller populations are allowed to stand in for or generalize to larger populations? For example, are findings for Danes claimed to represent Europeans? What about Sardinians or Amazigh?

Finally, sociologists of scientific knowledge have focused on the processes by which forms of “rationalization” are constructed to

build scientific authority. This study should encourage researchers to look at the roles of decidedly non-rational factors, like ambiguity, in the production of scientific authority and order, and to explore other roles of ambiguity. Under what circumstances do such factors become functional rather than disruptive? Such attention will help us develop more subtle accounts of the growth of scientific authority and rationalization.

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Data Note

Additional methodological information, including the data table for the content analysis, is available at <http://socgen.ucla.edu/people/aaron-panofsky/>.

Notes

1. This is true even for the “other” category, which we used mainly for religious labels and otherwise only in the very rare instances when a label did not fit one of the other classification systems.
2. In several instances, we coded terms that appear geographic as racial. For example, “Mexican American” because of the racialized way this term has historically been interpreted in the U.S. Census, and “East Asian” because this term is often used to distinguish “racial” Asians from Asian populations from the Indian subcontinent.
3. This shift does not seem to be due to overall growth of population labeling practices; animal research, for example, nearly always identifies the strain being used.
4. In 2008, the *Journal of the American Medical Association*’s deputy editor, Margaret Winker, presented data at the MIT “What’s the Use of Race?” conference, suggesting that the journal’s 2004 policy had not been fruitful.
5. One interviewee quoted earlier explicitly noted that highly specific labels help geneticists resist “inappropriate generalizations” of findings.

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