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Group Identity and Human Diversity: Keeping Biology Straight from Culture

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As the international effort to map the human genome matures, scientific interest in using that map to evaluate the genetic differences among human groups is growing (Collins et al. 1997). It recently has become popular (and politically important) to argue that this new interest in what might be called "population genomics" puts at risk significant interests of the groups under study and that those groups therefore should be involved in the decision to conduct any study that would use individual genotyping to generate information about the group as a whole (North American Regional Committee of the Human Genome Diversity Project 1995; Knoppers et al. 1996; Foster et al. 1997; Committee on Human Genome Diversity 1997). These proposals have vast practical and ethical implications. What would it mean for a group's collective permission to be "informed" and "voluntary"? If group consent is required, are other protections, such as the right to withdraw from research or confidentiality, also important for groups? How should the "researcher-group" relationship be managed administratively? Moreover, if the logic behind the argument for group rights in population-genomic research is accepted, it is likely to be applied to other biomedical spheres as well. Perhaps groups, and not just individuals, should have a say in whether they are included in genetic epidemiological surveys and preventive genetic-screening programs or whether customized treatments are developed to respond to their members' particular susceptibilities.

Before agreeing to impose new obligations on researchers, however, we need to give the propositions underlying the claims of group rights more scrutiny. Does population genomics put important interests of human groups at risk, and could some form of "group consent" succeed in protecting those interests?

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The Case for Group Rights in Genomic Research

It is important to note from the outset that there are real interests at stake for human groups in population-genomic research. For some studies, such as those in genetic epidemiology, there are risks analogous to those that individuals and families face in research aimed at identification of genes associated with disease. For example, finding that some particular group carries a relatively greater genetic propensity for alcoholism may be overinterpreted as universally predictive for the group (Edlin 1987). The resulting stigmatization of members of that group could then be used to deny social goods and opportunities to individuals who might be at no great risk of becoming alcoholics—or even to justify coercive and unnecessary medical treatments (Caplan 1994). As one of many unwelcome consequences to such a policy, the group's own sense of self-worth and solidarity might well be undermined.

Moreover, even the most academic population-genomic studies, such as the comparative genealogical efforts of the molecular anthropologists, can pose significant risks to groups. Consider this newspaper account from January 1996: "An extremely rare mutation on the Y chromosome may be a genetic marker that is unique to the people who first migrated to the Americas some 30,000 years ago, researchers report. . . . A group of Stanford University researchers have identified a mutation that in their sample (of 500 DNA samples from populations around the world) exists only in Indian populations in North and South American and in Eskimo groups. . . . The Y chromosome mutation occurred in a stretch of DNA that is not related to a gene, but is part of the 'junk' DNA that separates the genes" (Recer 1996).

Anthropologists report that some Native American groups already use the old racist doctrine of the "blood quantum" or the "One Drop Rule" as a way of defining membership in their social community (Moore 1996, p. 62). Would it be fair for an American man to use a positive test for this mutation in supporting a claim to affirmative-action benefits (see Caplan 1994)? What about women—and men with maternal lines of Native

American descent—who could not show this molecular "blood quantum"? Conversely, to the extent that anti-indigenous prejudice still animates the policies of some countries in this hemisphere, a detectable genetic hallmark such as this marker could serve as an indelible "yellow star," marking for oppression those with indigenous American ancestry. The capacity for both inclusive and exclusive uses of this little tidbit from the human-origin story already exists in genotyping labs throughout our hemisphere.

Troubles with Group Demarcation

I am persuaded by these arguments that human social groups do face significant risks from populational genomic research when they are used as proxies for human genetic populations. Nevertheless, I think that it would be a mistake for scientists and science policymakers to suggest that some process of "group consent" can protect these groups from those risks. What do we mean by "human groups" in the context of population genomics, anyway? If we mean genetic populations, or human "demes," we are not talking about the kind of human groups that can be approached for permission: they have no moral standing, deserve none, and, in any case, are unidentifiable until the research itself has been conducted. On the other hand, if we mean self-identified, morally authoritative social communities, approaching them for permission would be a hollow and dangerous gesture: by superimposing our social and biological categories, we would increase the risk of discrimination against the group members, and any protections that prior permission might afford would be undone immediately by both the modern human diaspora and the multiplicity of our group allegiances.

For the past 60 years, population geneticists have defined the groups that their science describes as "demes": groups of individuals more genetically similar to each other than to any other individuals (Gilmour and Gregor 1939). Most zoological population geneticists do not worry in advance about the boundaries of the different demes that they will find when they are assessing the patterns of genetic variation within a species in a given locale. They simply map out a grid over the terrain, collect and genotype random specimens from each square, and let the demic boundaries fall out of the resulting genetic-marker frequency data at whatever level of resolution suits their scientific purpose. Depending on the number and type of loci that they compare, those boundaries will shift, so that individual spiders sometimes can belong to one genetic population and sometimes to its neighbor. Since nothing much hangs on the boundaries of demes, except their scientific story, biologists are free to expand or contract their size as needed, to make their point (Wells and Richmond 1995). Indeed, not only are the boundaries between demes subject to revision as genetic data accumulate, but they also are intrinsically fuzzy. A glance through the maps found in wildlife field guides suffices to show that population ranges overlap, and careful observers are familiar with the prevalence of "intergrades" between subspecies. For this reason, drawing sharp divisions between clearly defined populations is usually an oversimplification of very complex data. A more accurate representation of phenotypic or genotypic data will be quantitative, not qualitative; it will rely on statistical measures such as a cline of distribution frequency, rather than on placing a boundary at some arbitrary locus.

The same is true in human population genetics. In theory, a survey of human genetic variation could be undertaken by use of ethnically anonymous blood samples collected randomly but systematically from blood banks, hospital drains, and battlefields around the globe. Given enough samples, a picture of human population structure could be developed, in which demes would be identifiable at multiple levels of resolution, from networks of people with specific unusual markers in common (such as particular germ-line *BRCA1* mutations) to the complex, fluid, but still largely endogamous populations (such as that of Central America) through which these networks are threaded (see National Research Council Committee on Human Genome Diversity 1997, p. 25). These demes would share the provisional nature and the blurred boundaries that are characteristic of the findings of field biologists.

With rare geographically isolated exceptions, however, the maps of human groups that would be produced by the field biologist's random-sampling approach to genetic diversity would bear little resemblance to a map of the world's self-identified autonomous human groups that are empowered to speak on behalf of their members. The population genomicists consistently have observed that "the basic conclusion from the study of differences among [self-identified social] groups is that they are small compared with the differences within the groups themselves. The aspiration of 'race purity' of classical racism is absurd. A village or a small tribe will show almost the same extent of genetic variation among individuals as will the whole world. Only human populations of very small islands that have been subjected for a long time to very close inbreeding show a moderate increase in genetic homogeneity" (Cavalli-Sforza 1993, p. 31).

Moreover, it is our membership in our socially constructed groupings, not our genetic membership in invisible demic families, that commands our loyalties, by giving us our connections, origin stories, and identities (Dominquez 1986). That is why it makes sense to point out that "within most indigenous epistemologies the questions of origins, the understanding of a people's

past, and early relations to other groups are dealt with in a very different way [than the scientific way]. Those communities already know where they came from, who they are and what their relations to the land are" (Moore 1996, p. 65). Lying unnoticed below the elaborate cultural constructions that unite and divide us, the impermanent and poorly defined human demes have neither the self-consciousness nor the moral standing to serve as guardians of their members' interests.

Furthermore, given our species' long history of using putative genetic relationships as the basis for nepotism, tribalism, racism, and aggression (Haller 1971; Marks 1995), aspiring to invest human demes with special moral standing seems wrong-headed in the first place. If we are right in our convictions that our biological roots should be irrelevant to the ways in which humans regard each other, promoting our genetic populations as groups with interests of their own makes no more sense than reviving old eugenic attempts to reify the concepts of "race," "genetic stock," or "germ plasm." Like those concepts, the genetic concept of a human "population" should provide only a way of organizing scientific data, not a way of classifying the human species.

Of course, the advocates of "group consent" for population-genomic studies do not have our silent and invisible demic connections in mind at all when they make their proposals; they are thinking of our loud and politically visible social networks. This is understandable. Human-population biologists are pulled by altruism (and pushed by politics) to focus their work on helping humans to improve their own welfare and self-understanding. That means answering questions framed in terms of the categories in which we already arrange ourselves (Marks 1995). Thus, it is the history, migration, and relative disease burden of humanity's many *socially defined* communities, not our anonymous demes, that drives most descriptions and defenses of population-genomic research.

Comparative genotyping and population genetics can still only try to tell those stories by measuring the flow and frequency of genetic markers. In order to do so, they seek markers and combinations of markers that will allow them to describe demes whose boundaries match as nearly as possible those of our culturally received ethnic groupings. Hence, this effort requires that DNA samples be collected from ethnically identified sources, and it is this methodological constraint that distinguishes the practice of many population genomicists from the straightforward, anonymous grid-sampling strategy of the wildlife biologists (see National Research Council Committee on Human Genome Diversity 1997). Strictly statistical analysis, yielding socially variegated demes such as "the population of Central America" would be an extremely inefficient way to reconstruct the groups that draw the population genomicists' interest. The rare Y-chromosome mutation marking indigenous American males is a significant demic demarcator not because it is something that most indigenous Americans have in common—to the contrary, it is quite rare—but, rather, because it seems to be sufficient to identify members of a group that we define through a particular origin story: the descendants of the "first migrants" to the Americas.

Population genomicists seem increasingly confident that, despite what we know about variation within groups, the polymorphic wealth of the genome will allow us to pick enough markers like this Y-chromosome polymorphism that are inherited in the same patterns as are our social identities, to serve as starting points for genomic studies (Barbujani 1997). On the other hand, these same scientists also believe that, as such studies proceed, the outlines of the two kinds of populations, ethnic and genetic, will probably begin to diverge and that the results of population genomic studies will affect our current beliefs about who we are.

The Trouble with "Group Approval"

If autonomous social groups are going to be asked to serve as the templates for genomic studies in this way, then the proponents of group approval seem to be correct: all the arguments in favor of respecting that autonomy and protecting their interests would appear to apply. But would the practice of obtaining approval achieve those goals? Such a process may, in fact, provide the control and protection that it promises, for many kinds of nongenetic epidemiological and clinical studies of the members of autonomous social groups (see Hall 1989). However, I do not think that this is true for population-genomic research, primarily because respecting their refusals to participate in such research would do nothing to protect them.

First of all, by constructing demes against the boundaries of real social groups and then reinterpreting those boundaries in terms of the demic results, this research suggests that the group's "real" identity is at the genetic level. As anthropologists have long pointed out about previous attempts to biologize social groups in order to undermine racist social policies, "the consequent confusion of biological and cultural characteristics, paradoxically, is the hallmark of racism" (Petersen 1980, p. 236). Reifying the deme, accepting the existence of objectively real and sharply defined biological groupings makes it easier for the group and its neighbors to stereotype its members (since they are now, by definition, "all alike" in some way) and to set them apart from the rest of humanity (since they are now, by definition, "different from" everyone else). No matter how great the potential of population genomics to show us our interconnections, if it begins by describing our differences,

then it inevitably will produce scientific wedges to hammer into the social cracks that already divide us.

Indeed, historically, the authority of the biological sciences often has been exploited whenever it has appeared to justify social disparities between groups, either by ranking groups hierarchically, using group-specific traits as stigmata, or rationalizing social barriers between groups (Haller 1971). Combating this kind of scientific racism has been the goal of heroic social-policy efforts during the past century. We should not risk undercutting that progress simply to take some shortcuts in our efforts to understand the dynamics of human evolution, and we certainly should not ask social groups to help us to do so at their own expense.

Second, the irony of asking social groups to serve as proxies for demes is that this cannot work to protect the communities' interests. First of all, no matter how careful researchers are to get permission from the right authorities in a local self-identified social group, as long as that group can be nested within a larger genetic population, its ability to protect itself from the consequences of the research will be compromised by any other subpopulation's decision to participate in that research. For example, despite that fact that many Native American groups have yet to be asked about participating in genetic-variation research involving Y-chromosome markers, they still must face the risks generated by the consent of the subjects in that Stanford University study, given its hemispheric level of resolution. Local community consent cannot give people control over findings at that level. Yet, as group-consent proponents acknowledge, populations at the level of "indigenous Americans" are never the kind of groups that one can reasonably approach for permission.

The problem of locating the appropriate "population" to consult is exacerbated even more by the increasing ability of human groups to scatter across the world and to be adopted by other culture. For example, imagine some quite–high-resolution genetic studies that would serve only to differentiate between the Hmong people and their neighbors in Southeast Asia. As long as the Americanized children of recent Hmong immigrants in Minnesota can be recruited successfully, seeking the consent of the Southeast Asian Hmong will not increase their control over their interests in this research. It would be odd to consider giving authorities in "the old country" the power to decide whether any Americans—Asian, African, or Irish—could participate in research that our own local institutional review boards have duly approved.

Ironically, in fact, the American melting pot threatens to vitiate the efforts of group-consent advocates to respect the autonomy of the rest of the world's peoples. In the wake of the controversy over the early descriptions of the Human Genome Diversity Project, the National Research Council Committee on Human Genome Diversity recommends that, as a show of good faith, American scientists begin their study of human diversity on our own population, so that we can absorb the brunt of whatever unanticipated risks may occur. Moreover, they reason that "as a result of centuries of immigration, the United States does have a diverse representation of the people of the world. Thus, a well designed survey of human genetic variability there could shed some light on the extent of human genetic variation globally" (National Research Council Committee on Human Genome Diversity, p. 73).

To the extent that this idea's scientific benefits are well founded, however, its merits as an ethical gesture are dubious if one accepts the claims of the group-consent advocates: if we are the world, then, by studying ourselves, we will be studying the world's social groups, without any consent at all, and involuntarily will be imposing on them all the risks of stigmatization, discrimination, reductionism, and self-alienation that their cohorts in our country voluntarily assume.

Summary and Next Steps

To the extent that population genomicists seek to use self-identified, politically organized groups in their research, even as phenotypic placeholders, these groups should be given the same panoply of rights that we give to individual human subjects in biomedical research: free and informed agreements to participate, withdrawal rights, confidentiality protections, control over the disclosure of identifiable research results, and just compensation. Allowing groups to exercise those rights, of course, would kill many population-genomic studies in their cradles. But that is not why it would be a bad idea to involve social groups as gatekeepers for population-genomic research. It would be a bad idea for two main reasons: First, it would send the wrong message, by suggesting that geneticists think that there really is a strong biological justification for the social boundaries that we draw around and between each other. That flies in the face of the scientific observation that human groups that will be picked out, described, and compared in the course of population-genomic research are almost always the results of mixed lineages that make hash of most of our familial-origin stories and social groupings. Demes are not autonomous, self-identified human groups, and it would be dangerous to devise a system that suggested that particular social groups could speak for them. Second, obtaining group approval would not give groups control over research risks. Both our practice of nesting local groups within larger social categories and the prevalence of cross-cultural immigration mean that no one group with the voice to do so can have the

moral reach to decide for all those who might participate in such research.

This argument suggests that it is hollow rhetoric to promise groups that, through a process of group approval, they will have the power to protect themselves from the risks of population-genomic research. But, if that is the case, how should we proceed?

First, we should look for scientific alternatives to the use of identified social groups as templates for population-genomic research. One of the revolutions that has spurred the recent progress of molecular genomics has been the advent of "positional cloning" as a gene-hunting strategy. It has been revolutionary, because it has reversed the usual starting point of a genetic study, by proceeding from the the molecular genotype to the clinical phenotype. A similar revolution is now required for comparative population genomics. If methods for the assessment of genetic diversity can be developed that do not have to start with the "phenotypes" of the social landscape, then perhaps studies can be designed that, like those of field biologists, allow demic boundaries or the representations of population clines to be drawn as the available human genetic data permit. If the results of such studies are unwelcome to some social groups (as they are bound to be, in some cases), geneticists still can insist that they owe no allegiance to our various cultural myths but simply must be true to their own tools.

Finally, perhaps our policies should make it clear that it may never be possible to protect groups such as "Irish Americans," "African Americans," and "Native Americans"—groups that are too abstract and heterogeneous to have a voice but whose self-identified members still can suffer from the labels that they acquire. At the very least, however, we should work to make it more possible for individuals to consider the interests of other members of such groups, as part of their own informed-consent decision making. If potential DNA donors were informed of the risks that their donation would impose on all others who share their broadest social identities (and not just on their local community), they could incorporate those collateral risks into their decisions, just as individuals now incorporate the interests of their nuclear families into decisions to pursue clinical genetic testing. Participants in population-genomic studies—both the scientists and their subjects—need to be reminded, above all, that our moral communities are our cultural households, not our genetic connections, even when we mistakenly build the former on our perceptions of the latter.

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