

Human Genome Diversity Studies: Impact on Indigenous Communities

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The study of genetic variation in the human species cannot be undertaken outside the cultural, political and historical context within which scientific research has been traditionally conducted on indigenous peoples. Consideration of their rights rendered the Human Genome Diversity Project intractable in the 1990s, and most issues remain unresolved.

Introduction

The Human Genome Diversity Project (HGDP; Cavalli-Sforza *et al.*, 1991) was proposed as an augmentation to the Human Genome Project (HGP). Recognizing that the HGP was based on a Platonic design in which the human species was represented by a single ideal specimen, population geneticists proposed the collection of genetic material from diverse populations of the world. The project came under intense scrutiny and criticism, and was ultimately abandoned. It has now been replaced by the Geographic Project (GP), which comes with private funding.

Why the Focus on Indigenous Communities at All?

Many of the problems faced by the HGDP were brought about by its insistence on targeting indigenous, exotic peoples rather than urban populations. If the goal was to study the human gene pool at the beginning of the twenty-first century, it was flawed, because the indigenous peoples only represent a small fraction of that gene pool. The project could be better served, notes the National Research Council's report from 1997, by collecting samples from internally diverse urban populations.

The HGDP was structured around a single research topic: the microphylogeny of the human species – the pattern of descent of different human groups – which had been a principal research issue of the HGDP's leader, Stanford

geneticist Luca Cavalli-Sforza. Using statistical analyses of the frequencies of many alleles across many populations, Cavalli-Sforza represented the similarity of human gene pools in a tree-like structure or dendrogram, which resembled the ancestry of species. But these dendrograms grossly oversimplified the historical processes affecting human populations. As a result, these depictions were unstable and were sensitive to the statistics used, the genes analysed, the particular populations chosen and the demographic history of the groups.

If the HGDP had been principally interested in the structure of the contemporary human gene pool, it might have begun by sampling according to an arbitrary criterion such as geography, as one of its principal organizers, Allan Wilson, suggested at the outset. But its guiding question was the genetic relations among culturally designated groups, and so it adopted the cultural groups themselves as its organizing principle for sampling, which became the root of the controversy.

Historical and Political Context

Blood has been retrieved in the field by anthropologists since the development of serological technology early in the twentieth century. Carleton Coon was the first to retrieve blood (in 1922) from 'his people', the Rif, in Morocco, to see whether their physical features and their blood-group features would match when allocating them racially.

Blood remained a staple of anthropological collection even as the questions changed and race waned as the dominant anthropological paradigm. Even so, it had special problems associated with it, as Coon himself noted in the 1950s:

Blood-letting for blood-group analysis falls into the class of blood-letting in general, and evokes the whole ideology of blood-brotherhood, the fear of injury by contagious magic, and that of ritual contamination based on the analogy of menstruation. (Coon, 1954)

Advanced article

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Blood is never, as anthropologists have been known to say, 'just' blood. Nevertheless, its collection had proceeded for decades on a small scale, sometimes as part of a specifically anthropological project or sometimes for medical testing, and it has been retained and 'piggybacked' by researchers interested in other questions. The overarching assumption here has been that once the substance is out of the person's vein, it belongs to the researcher.

This followed tradition in classical anthropology: the great collections of Native American skeletal materials, for example, were 'acquired' in the nineteenth and twentieth centuries through practices that even included grave-robbing. 'It is most unpleasant work to steal bones from a grave', wrote the great anthropologist Franz Boas to his sister very early in his career, 'but someone has to do it'. Ultimately, the bones came under the control of museum scientists and were used to advance the careers of the many scholars who acquired them or analysed them, often without regard for the sensibilities of the people whose relatives the bones actually comprised. It was a classic colonial enterprise, agents of a powerful state acting with little regard for the powerless.

The situation changed dramatically with the passage of the Native American Graves Protection and Repatriation Act of 1990 (NAGPRA). This legislation was designed to acknowledge that North American Indian remains were sacred objects, no less sacred than the scientists' relatives' bones and belonged not to science but to the tribes from whom they had been 'acquired'. This was a significant affirmation of the rights of indigenous Americans and came at precisely the same time that the HGDP was being formulated and was naively planning to collect bioanthropological objects of sacred value on a large scale.

A second significant political context involved widespread rumours within indigenous communities, who were finally beginning to get attention of 'white people' plotting to steal the body parts, bodies or simply blood of indigenous people. In some cases they were right, as an international trade in body organs later developed.

A third political context involved the development of 'biocolonialism' by agribusiness. Availing themselves of 'knowledge' freely given by indigenous people, large agricultural corporations were making considerable profits in which the people whose knowledge they needed were not sharing. In addition, patent law concerning biotechnology strongly favours scientists, as the unsuccessful cases of John Moore (Greely, 1998) and the descendants of Henrietta Lacks (Jackson, 2001) showed – neither was permitted to share in the profits made from cell lines derived from their bodies. In the case of the blood of indigenous people, the National Institutes of Health applied for patents for cell lines derived ultimately from the blood of a Hagahai (Papua New Guinea), a Solomon Islander and a Guaymí (Panama). This seems to recreate the scenario of the rapacious North American capitalist, looting not merely the land or artefacts or knowledge of tribal people, but now their very blood.

Group Consent

Since the HGDP was conceptualized around human groups, each individual sample is only interesting to the extent that it is a representative of that group. Consequently, the idea of 'group consent' was devised as a means of securing not only the voluntary participation of the particular blood donor, but also the voluntary participation of the polity represented by the donor.

While introducing group consent was an admirable step in principle, it raised a significant number of ancillary issues, centring on the idea of representation: which blood samples represent which peoples? After all, human groups are fluid and organized hierarchically. Is group consent relevant to someone who is a 'Chiricahua Apache', an 'Apache', an 'Athapaskan' and an 'American Indian' simultaneously? If the Northern Paiutes decide not to participate in a study and the Southern Paiutes decide to participate as Paiute representatives, are the rights of the Northern Paiutes thereby violated? If the Hopis decline to participate in a genetic study, can geneticists be prevented from soliciting samples from acculturated Hopis living outside their reservation?

Further, the solicitation of permission from a political entity representing the people raises the problem of possible coercion. If the leaders agree to participate, does an individual still have free rein to refuse, or can the leaders now simply act as agents for the scientists and subtly compel compliance? And more specifically, does everyone in the decision-making complex understand fully what the scientists want, why they want it and what they plan to do? For people who do not share scientific comprehensions of blood, cells, deoxyribonucleic acid (DNA), identity, life, illness and medical genetics, the elicitation of full informed consent would seem to necessitate the development of a crash course in local biological idioms.

Ultimately, the issue of group consent would also serve to reify these groups genetically as units of nature, when they are in fact units of social, political and historical manufacture. Perhaps the best statement about the problem of relating human group-assignment to the reality of identity is given by the late Frank Dukepoo (1998, p. 242), a Native American geneticist:

I call myself a 'full-blood' American Indian of Hopi and Laguna heritage. While constructing my own pedigree, I found this is far from the truth: my father (a 'Hopi') is a mixture of Hopi, Ute, Paiute, Tewa and Navajo; my mother, on the other hand, (a 'Laguna') is a mixture of Laguna, Acoma, Isleta, Zuni and Spanish. Members of other tribes share similar admixture histories as our ancestors raided, traded or kidnapped to ensure survival of their numbers. As it is reasonably safe to surmise the same situation for members of other ethnic groups, what would 'diversity' research reveal?

Pragmatic Concerns

In an early attempt to muster interest for the HGDP, its advocates used arguments from 'salvage anthropology' – the impending loss of these peoples, an argument familiar to anthropologists since the middle of the nineteenth century. However, it sounds very cynical to ask for blood from people who are on the brink of extinction. A native of the Solomon Islands wrote:

The project has very little interest in helping these people to survive, or in addressing the social, the economical, the political, and the exploitation issues that endanger these indigenous groups of people. (Liloqula, 1996)

Moreover, some groups were simply experiencing the normal historic forces of merging, splitting, reconstituting and forging new identities; they were 'endangered' only as bounded genetic entities.

Participation in the benefits of modern healthcare often requires allowing blood to be drawn. Some insecurity naturally arises about the fate of that blood – a highly symbolically charged substance – once its diagnostic purpose has been served. Many tribal people would feel very uneasy to learn that their blood, or a product derived from it, was sitting in a laboratory in California, and being manipulated exclusively for the benefit of American scientists.

On one occasion, when Cavalli-Sforza was taking blood from schoolchildren in a rural region of the Central African Republic, he was confronted by an angry farmer brandishing an ax. Recalls the scientist, 'I remember him saying, "If you take the blood of the children, I'll take yours"'. He was worried that we might want to do some magic with the blood.' (Subramanian, 1995, p. 54)

Rather than musing over the ignorance of the ax-wielding farmer, a contemporary reader should instead recognize unfulfilled obligations of disclosure to the participants here. A strong fear of the magic in blood makes it very unlikely that these people could have given their fully informed consent to this research.

The existence of such ideologies about the power of blood could in principle be circumvented by stipulating that the blood, once drawn, can be used only for the medical purpose specified and must then be destroyed. That is commonly the case now with some North American Indian groups, but there is unfortunately no way to enforce it. Blood from indigenous peoples has been a valuable scientific commodity, traded between laboratories and researchers, for different projects, establishing a network of relationships, obligations and coauthorships (Anderson, 2000). The prospect of such a scientific tradition abruptly ceasing is quite unlikely, regardless of the agreements made with tribes, who may also be distrustful of Euroamerican people living up to their agreements at all.

The question of genetic exploitation is of paramount importance. If there is economic value in the blood of

indigenous people (as the interest of biotechnology companies might suggest), then what is a fair price? The HGDP's insistence that there are no financial considerations was forcefully undermined by the patent applications, which did not involve the HGDP itself but were obviously relevant. Consequently, any adequate concept of disclosure and voluntary informed consent would necessitate the scientist explaining to participants that there are financial stakes, through which the researcher could get wealthy without precedent for the subject sharing in that wealth!

Since the HGDP's initial interest was to formulate and answer questions of microevolution, another issue is raised, calling attention to science's role in authoritatively contradicting people's ideas of their folk history and identity. One could legitimately ask why anyone would wish to participate in a project designed to undermine their own ideas of who they are and where they came from. Indeed, the Havasupai tribe in northern Arizona agreed to give blood samples to researchers specifically for a study of diabetes, and learnt some years later that samples were being used for studies of the peopling of the New World. Believing that they are autochthonous, however, the Havasupai maintain that they would not have agreed to participate if that aspect of the research has been presented to them.

In any case, medical value, which came to be emphasized a few years after the HGDP's inception, would be difficult to establish, as no plans were made for collecting detailed medical, phenotype and life-history data to associate with the genotypes. A study of the genetic aetiology of diabetes, for example, would require a knowledge of which DNA samples actually came from people who were diabetic. However, those samples could then not be used for a study of the genetics of schizophrenia, because there would be no information about which samples came from schizophrenics. Thus the HGDP samples could only have been of exceedingly limited medical use.

Into the Twenty-first Century: The Private Sector

Unable to grapple successfully with the diverse issues it had raised, the HGDP abandoned its hope to secure funding from the US government. In 2005, it was succeeded by the GP, underwritten by National Geographic, IBM and the Waitt Family Foundation (founders of Gateway computers). Its explicit goal is that of the HGDP – elucidating the microphylogeny of human populations, and it makes no claims to any other ambitions. Also like the HGDP, however, it was formulated without input from bioethicists; but with private funding it faces fewer constraints than did the HGDP (Annas, 2006). While targeting indigenous peoples, the GP nevertheless tries to engage those of the urban, industrialized world by allowing them to submit their own samples, for a charge of \$99.

Acknowledging the apprehension its lack of bioethical oversight might produce, the GP attempted to demonstrate

its good will by indicating that it would utilize the Institutional Review Boards (IRBs) from the universities at which its scientists are employed. A problem quickly arose when The University of Pennsylvania's IRB approved the collection of DNA samples from Alaskan natives, pending the approval of a local Alaskan IRB. The samples were collected, however, without such approval. The result is that the GP is presently negotiating to retain the samples, which presumably should not have been collected in the first place (Harmon, 2006). Faced with the informed cynicism of various Native American groups (Schroeder *et al.*, 2006; Marks and Harry, 2006), the GP has shifted its focus instead to the world's other indigenous populations.

Venture capital has transformed human population genetics in other ways. Various companies now offer services that examine mitochondrial or Y-chromosome DNA sequences, and use them to link clients to particular indigenous peoples (Wald, 2006). Generally, these DNA sequences are widespread and polymorphic, the indigenous populations themselves are poorly sampled, and the manner in which the samples were collected remains largely unknown. The establishment and exploitation of such fictive kin ties on the basis of an uncritical acceptance of genetic data has come to be known as 'biosociality'.

Finally, the question of the natural divisions of the human species has been resurrected, through the introduction of pharmaceuticals targeted at particular 'races'. Paradoxically, in a last-ditch effort to gain support, the HGDP had avowed it would delegitimize group hatreds by demonstrating the nonexistence of race. A few years later, however, genetically reified races now present the possibility of 'niche marketing' and the creation of new sources of revenue for big pharma, despite being contradicted by known patterns of human biological variation – which are principally polymorphic, clinal and local.

Ultimately, the impact of the human genetic diversity studies has been to reinforce many communities' worst fears about the avarice of wealthy nations and the residual colonial attitudes of science, appearing more as an instrument for their exploitation than as a fulfillment of the Baconian promise of a better life for all. **See also:** [Community Consent for Genetic Research](#); [Human Genetic Diversity](#); [Human Genome Diversity Project \(HGDP\)](#)

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Further Reading