

RACE AND GENE STUDIES: What Difference Makes a Difference?

In 1851 the Louisiana physician Dr. Samuel Cartwright observed a behavior evident in African Americans but absent in whites. They tended to run away from slave plantations. He attributed this odd behavior to a disease peculiar to Negroes. He even gave the affliction a name, "drapetomania."

Cartwright's "run-away" disease elicits derisive laughter today. So too do all the other 19th and early 20th century exertions to distinguish races by facial angle, skull size, cranial index, length of shin bone and blood type. Our growing knowledge of the genome and human evolutionary history help us understand why all such efforts to locate the source of innate racial difference were doomed: it doesn't exist. Most geneticists and anthropologists who study human variation agree that humans just don't come bundled into three or four separate groups according to skin color and other physical traits.

Nonetheless, "discoveries" of racial difference resurface in the press with predictable regularity. Often they spotlight differential rates of diseases, or responses to a drug. Sometimes they zero in on a genetics study.

That was the case recently with a report published in Science magazine (Dec. 20, 2002). Noah Rosenberg, Marcus Feldman and others analyzed the variation in 377 different DNA sequences from 1056 individuals from around the world. They found that 95% of the DNA variation they studied is due to differences between individuals within any continent. But they also found they could use the remaining 5% of the variation as genetic "footprints" indicating the continent from which an individual's recent ancestors came.

Some were quick to interpret these results as evidence that old-fashioned notions of the "races of man" have been correct all along. But does it? What do these studies actually tell us? And why should our interpretations matter?

Reports about such studies commonly fall prey to three confusions: they conflate DNA markers of ancestry with markers of race. They mistake the fact that some gene variants are more common in some populations than others as signs of racial "difference" between those populations. And they assume that disparities in group outcomes can be attributed to inborn, or genetic, differences between races.

The idea of biological race assumes traits come packaged together, even color-coded for our convenience, as anthropologist Jonathan Marks jokes. In other words, if biological race were real, we'd find that skin color or other "racial" markers would correlate with a suite of other genetic traits. Knowing an individual's "race" should enable us to predict his or her other genes and traits.

But the DNA sequences studied by Rosenberg and his colleagues are not genes. Known by geneticists as "microsatellite short tandem repeats" (and more colloquially as "junk DNA"), they do

not code for proteins, but just sit there taking up space in our DNA. Mutations in DNA sequences that don't code for anything are not affected by natural and sexual selection. They are neither selected for nor against but are simply passed down, generation to generation. Comparing these accumulated mutation patterns can provide clues to ancient population movements. But they have no effect on physical traits such as skin color or hair form or blood type.

In other words, the study accomplished the same thing our eyes do everyday. You can look at someone and stand a pretty good chance of identifying the continent where that person's recent ancestors lived, especially if you're gazing at someone whose family has resided in the same place for several generations - as did all the subjects of the study.

But what's that got to do with "race"? We all have ancestors from elsewhere - and if we go back far enough, about 70,000 or so years ago, all our ancestors can be traced back to Africa. But if our idea of race assumes that different groups each share among themselves a different suite of inborn traits, then we have to ask, "What difference makes a difference?" Certainly not micro-satellite short tandem repeats.

Still, there's no question that some gene forms show up more often in some populations than others: alleles that code for blue eyes, or the A, B, O blood groups, and of course, those alleles that influence skin color. (We all have the same 20,000 or so genes. But some genes come in different forms, or varieties, called alleles.) But just because some members of a population might carry a specific gene form, doesn't mean all members do. Only a small percentage of Ashkenazi Jews carry the Tay-Sachs allele. When a couple I know were screened upon their pregnancy, the non-Jewish partner was found to be the Tay-Sachs carrier, not the Jewish one.

That's because most human variation falls within, not between populations. About 85% of all genetic variation can, on average, be found within any local population, be they Swedes, Kikuyu, or Hmong. About 94% can be found within any continental population, consistent with what the Rosenberg Science study found. In fact, there are no characteristics, no traits, not even one gene that turns up in all members of one so-called race yet is absent from others.

Take sickle cell. Doctors were long taught that sickle cell anemia was a genetic disease of Negroes, a marker of their race. Yet sickle cell is found among peoples from central and western Africa, but not southern Africa. It is also carried by Turks, Yemenis, Indians, Greeks, and Sicilians. That's because sickle cell arose several thousand years ago as a mutation in one of the genes that codes for hemoglobin. The mutation soon spread to successive populations along the trade routes where malaria was common. It turns out that inheriting one sickle cell allele confers resistance to malaria and thus provides a selective advantage in malarial regions (inheriting sickle cell alleles from both parents causes sickle-cell disease). In other words, sickle cell, like tandem repeats in the Science study, is a marker not of skin color or race but ancestry, or more precisely, having ancestors from where malaria was common.

Like sickle cell, most traits are influenced by separate genes and inherited independently one from another. They are said to be "non-concordant." Someone with brown hair might carry A, B or O

blood. Sub-Saharan Africans tend to have dark skin. But so too do Dravidians from India, Aborigines from Australia, and Melanesians from the South Pacific. Large numbers of West Africans are lactose intolerant as are Japanese, but East Africans aren't. German and Papua New Guinean populations have almost exactly the same frequencies of A, B and O blood. At one point on the genome an individual might share a gene form common in Africa, at another site East Asia, and still another, Europe. Jared Diamond and others have pointed out that for each trait we can classify people into "races" by that trait, each giving us different and overlapping races depending on the trait selected.

Indeed, the Rosenberg team found they could cluster the individuals in their sample into several different statistically significant groups, only one of which corresponded to five continents. They also found that no matter which clustering scheme they used, individuals could be placed in more than one group.

The reason for all this within-group variation is because unlike most other species, modern humans, *Homo sapien sapiens*, are young, only about 150,000 years or so old, and we've always moved. Human populations just haven't been isolated from each other long enough to evolve into separate sub-species, or races. As humans migrated around the globe, populations bumped into each other and shared their mates - and genes. Sometimes genes flowed across great distances - through trade, war, slavery, piracy, exile and migration. More often they flowed from village to village to village.

Ancestry is important in genetics and health care. I would like to know if I were descended from a population at risk for sickle cell, or the blood disease porphyria, or Tay Sachs. Oliver Sachs has even written famously of an island of the colorblind. But because of non-concordance, the geographic shape of populations at genetic risk for a specific disease or adverse drug response change amoeba-like depending upon the trait under study. These populations do not map onto what we think of as race. To assume they do is medically troublesome on several accounts.

First, doctors might be tempted to use race as a very unreliable surrogate for an individual's own unique ancestry and patterns of inheritance. And insofar as 94% of all genetic variants can be found within any continent, assigning someone to a "racial" continent of origin is too gross a scale to narrow down the range of possibilities very much. (Some variants do correlate on a finer scale with ethnic groups, and on a scale smaller yet with recent family descent.)

But a belief in biological race also obscures the very salient consequences of race as lived experience. Race may be a biological myth, a social construction, but it nonetheless remains very real. It can even have biological effects. African Americans have among the highest rates of hypertension in the world. This was long assumed to be genetic, a "marker" of their nature. But then it was found that West Africans have among the world's lowest hypertension rates. A focus on race as innate biology, as genetic difference, would lead health professionals and policy makers to overlook social factors that might contribute to African American hypertension and heart disease, including the added stressor of living in a racist society.

Race is terribly relevant to life outcomes. The likelihood that toxic waste has been dumped in your neighborhood, your ability to get a home loan, the quality of your kid's education, connections to job opportunities, whether or not you're likely to be followed in a department store or pulled over by police, are all influenced by your race. Race does matter. Not race as genetics but race as lived experience, what sociologists call "social" race. Social race is an important variable for health researchers and epidemiologists.

The factors that lead to differential outcomes between races live not in any "racial" genes but in our social institutions and practices. It's easy to confuse the two. But doing so, like Dr. Cartwright's drapetomania, displaces our attention from those discriminatory practices to the "nature" of the victims. Blindness to the continuing impact of racism can be just as harmful as believing that race is biologically real. They both let society off the hook.

By Larry Adelman, Executive Producer, ***Race - The Power of an Illusion***

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