Revisiting Race in a Genomic Age

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At the beginning of modern human genetics, the classical book by Neel and Schull devoted half a page to "race" with the explanation that "in the past the term has been used in so many senses by so many writers that it has largely lost its usefulness."[1] In the next half century, there were a few explorations of race in genetic epidemiology and population genetics, but discussion was omitted from most texts until it became indispensible for evaluation of forensic DNA evidence. The conflict between geneticists who value racial differences and anthropologists who dismiss them increased the first group's profits by developments that the second rejects. I will return to this conflict after reviewing a new book that addresses the argument, unfortunately before important developments in the past two years.

In brief, the text is divided subequally into four sections dealing with concepts of race, race-targeted research, genetic ancestry, and race in public discourse. Within each part there is at least one author sympathetic to race and at least one strongly opposed for philosophical or other reasons. The first section deals with the long history of the race concept and its interactions in the genomic age with different sciences and applications to markets and medicine. The interdisciplinary dialog is fascinating, with different associations attached to race, ethnicity, nationality, group, population, affinity, ancestry, and similar terms that for me were not clarified without consulting other sources. In the second section, Feldman and Lewontin address race and ancestry, adopting for illustration a procedure excluding interracial crosses that Balnick politely but strongly criticized in the previous article. They clarify this in their last section and conclude that "confusion between race and ancestry …. is critical and must be accounted for in medical practice …. For diagnosis and treatment, however, individual genotypes will, in the long run, provide the most useful information." Tate and Goldstein cautiously note that "some of the variation in how medicines work may correlate with 'racial' or 'ethnic' groups, exacerbating health disparities." Their chapter is outstandingly even-tempered. Kahn makes the clearest presentation of BiDil and other drugs that are prescribed for a race-specific group rather than an identified genotype, a policy that favors patents and drug approval. Racial patents are bound to increase under current U.S. law, if, as the Royal Society argues, the promise of truly individualized pharmacogenomic therapies remains decades away. The next two chapters document problems that arose when the NIH launched programs that demanded racial categories for which the data were minimal and therefore the assignment of mixed races was controversial.

The third section extends this problem to genetic ancestry inferred, mostly from mitochondrial DNA and nonrecombing part of the Y chromosomes. Applied to African Americans and other multiethnic individuals with little pedigree information, the reliability of inferred genetic ancestry is uncertain and decreases as the inferred origin is more specific. The exercise (carefully described by Shriver and Kittles) becomes in less critical hands a triumph of commerce over science. Greely describes similar problems for racial inference in genetic genealogy, whereas Tallbear introduces the DNA testing of individuals who want membership in a native American tribe for either government entitlements or emotional satisfaction. Finally, Nelson reviews the social forces that encourage members of a diaspora to seek their roots by genetic genealogy testing.

The last section has the greatest diversity between the recent argument of Lee and the intensity of Stevens, who wants geneticists to be supervised by nonscientists assigned to guarantee that race would not be mentioned in a manuscript submitted for publication. The justification of this policy is "the self-fulfilling prophesy of genetic studies, many funded by the NIH, that assume but do not prove that races are genetically discrete." Fortunately there is encouraging counter evidence. The first is a paper by Neil Risch et al. in favor of self-identified race and ethnicity.[2] At a time when genotyping was in its infancy, they proposed that "race" be applied to five major groups (African, Caucasian, Asian, Pacific Islander, and native American), with "ethnicity" reserved for subgroups of the above. This conforms to *A Dictionary of Epidemiology*,[3] but does not allow for racial admixture, for which they suggested *ancestry* as a combination of 2–3 races or ethnic groups. A recent paper by Chakravarti is equally supportive of race, although cautious about the causes of common diseases and proposes to explain ancestry by whole-genome analysis.[4] This is a reasonable objective, but far from immediate worldwide solution.

Between these past and future extremes is the success of the Wellcome Trust project to detect causal genes for complex diseases by using whole-genome scans of cases and controls.[5] In the years between 2007 and 2008, diseases that had defied linkage analysis provided strong support for up to 30 causal genes in a defined racial

*Correspondence: nem@soton.ac.uk
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sample.[6] It was no longer defensible to argue that race is meaningless, although it remained possible to speculate that some unspecified subsamples might be biased by environmental effects. This is an incredibly difficult hypothesis to test convincingly, because there is no information to test for the effects of unspecified but possibly relevant environments. Enthusiasts of environmental differences can no more prove their hypothesis than geneticists can at present disprove it. This deserves more attention than it receives, which may not come until the genetic and environmental evidence are both exhausted. Meanwhile, genetics is advancing and anthropology isn't.

At this point it is tempting to hypothesize about how this conflict will end, most likely by retreat of the anthropologists and perhaps by discovery some time in the future that a small proportion of genome claims are misinterpretations of environmental differences. Epidemiologists are more favorable to this hypothesis than geneticists, leaving genetic epidemiologists in a quandary that can be solved only by evidence.

Meanwhile, experience since the beginning of genetic epidemiology seems relevant. From a Marxist position, Lewontin[7] argued that zero heritability of socially important traits like intelligence cannot be excluded on present evidence. Some effort was devoted to testing this hypothesis with the conclusion that heritability may well be less than conventional estimates, but no model yet invented is consistent with zero heritability.[8] Analysis could be pushed further. William Shockley, inventor of the transistor, Nobel Laureate, and member of the National Academy of Sciences, in his last years argued that African descent is inextricably associated with a 15% decline in the intelligence quotient, as James Watson asserted much later. That was a serious error, contradicted unequivocally by analysis of the earlier data[9] and current evidence. There is no justification for imagining an effect contradicted in these ways and supported by none. Given sufficient evidence, genetic epidemiology can resolve controversy generated by prejudice from either political extreme.

To universal amazement, this issue was raised again when Rose argued that scientists should not study race and IQ because the information gained does not "contribute to basic scientific understanding, offer new beneficial technological prospects, or aid scientific policy making."[10] On the contrary, the evidence cited above contributes far more to society than a Marxist denial of science, a point made with three recent supporting studies and the rebuttal of Rose by Ceci and Williams.[11]

This book might be very different if it absorbed recent evidence that provides the "critical first step toward interdisciplinary dialogue" promised in the introduction. How anthropology will adjust to the current success of human genetics is a question that lies outside our remit, but it has potential to help or harm an active science. By covering problems ranging from serious to imaginary (but dangerous), this book should be required reading for anyone concerned about the future of human genetics in the U.S.

References

1. Neel, J.V., and Schull, W.J. (1954). Human Heredity (Chicago: University of Chicago Press).
2. Risch, N., Burchard, E., Ziv, E., and Tang, H. (2002). Categorization of humans in biomedical research: genes, race, and disease. Genome Biol. *3*, 1–12.
3. Last, J.W. (1995). A Dictionary of Epidemiology, Third Edition (New York: Oxford University Press).
4. Chakravarti, A. (2009). Being human: Kinship: Race relations. Nature *457*, 380–381.
5. Wellcome Trust Case-Control Consortium. (2007). Genome-wide association study of 14,000 cases of seven common diseases and 3,000 shared controls. Nature *447*, 661–678.
6. Barrett, J.C., Hansoul, S., Nicolae, D.L., Cho, J.H., Duerr, R.H., Rioux, J.D., Brant, S.R., Silverberg, M.S., Taylor, K.D., Barmada, M.M., et al. (2008). Genome-wide association defines more than 30 susceptibility loci for Crohn's disease. Nat. Genet. *40*, 955–960.
7. Lewontin, R.C. (1975). Genetic aspects of intelligence. Annu. Rev. Genet. *9*, 387–405.
8. Rao, D.C., Morton, N.E., Lalouel, J.M., and Lew, R. (1982). Path analysis under generalized assortative mating. II American IQ. Genet. Res. *39*, 187–198.
9. Rao, D.C., Morton, N.E., Elston, R.C., and Yee, S. (1977). Causal analysis of academic performance. Behav. Genet. *7*, 147–159.
10. Rose, S. (2009). Should scientists study race and IQ? No: science and society do not benefit. Nature *457*, 786–788.
11. Ceci, S., and Williams, W.M. (2009). Should scientists study race and IQ? Yes: the scientific truth must be pursued. Nature *457*, 788–789.

Newton E. Morton[1,*]
[1]Faculty of Medicine, Department of Human Genetics, Duthie Building (MP 808), University of Southampton, Southampton SO16 6YD, UK