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Scientific Outsiders and the Human Genome Project. Review of Timothy F. Murphy and Marc A. Lappé, *Justice and the Human Genome Project*; Robert F. Weir, Susan S. Lawrence, and Evan Fales, *Genes and Human Self-Knowledge: Historical and Philosophical Reflections on Modern Genetics*; Tom Wilkie, *Perilous Knowledge: The Human Genome Project and Its Implications*

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Abstract

The Human Genome Project, the international effort to map and sequence the genetic material of *Homo sapiens*, has by now generated a mass of information about DNA sequences. It has also generated an independent, but related, mass of texts exploring the philosophical, historical, sociological, and legal implications for medical care, human identity, law, politics, and reproduction that the project raises. Indeed, the Human Genome Project is perhaps most noteworthy for its status as the first and only scientific project to fund independent studies of its own social implications. The genome project budget in the United States, which is divided among several federal agencies including the National Institutes of Health and the Department of Energy, includes a generous amount set aside for bioethicists, policy planners, historians, philosophers, and other scholars. Of the three books reviewed here, only one (*Justice and the Human Genome Project*) has any connection to this funding mechanism. But all reflect the popular and political interest that the genome project has provoked. Few of those commenting on the genome project in these studies are laboratory molecular biologists familiar with polymerase chain reaction, in situ hybridization, or any of the other technologies for manipulating DNA that have been so important to the project. They are, instead, scientific outsiders who are expected to shed light on the long-term social implications of the access to hereditary information that the genome project promises to make possible.

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Scientific outsiders and the Human Genome Project

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Justice and the Human Genome Project
Edited by Timothy F. Murphy and Marc A. Lappé
Berkeley and Los Angeles: University of California Press, 1994
200 pp. Clothbound, $28

Genes and Human Self-Knowledge: Historical and Philosophical Reflections on Modern Genetics
Edited by Robert F. Weir, Susan S. Lawrence, and Evan Fales
Iowa City: University of Iowa Press, 1994
268 pp. Clothbound, $29.95; paperback, $14.95

Perilous Knowledge: The Human Genome Project and its Implications
By Tom Wilkie
Berkeley and Los Angeles: University of California Press, 1994
202 pp. Clothbound, $20

The Human Genome Project, the international effort to map and sequence the genetic material of Homo sapiens, has by now generated a mass of information about DNA sequences. It has also generated an independent, but related, mass of texts exploring the philosophical, historical, sociological, and legal implications for medical care, human identity, law, politics, and reproduction that the project raises. Indeed, the Human Genome Project is perhaps most noteworthy for its status as the first and only scientific project to fund independent studies of its own social implications. The genome project budget in the United States, which is divided among several federal agencies including the National Institutes of Health and the Department of Energy, includes a generous amount set aside for bioethicists, policy planners, historians, philosophers, and other scholars. Of the three books reviewed here, only one *(Justice and the Human Genome Project)* has any connection to this funding mechanism. But all reflect the popular and political interest that the genome project has provoked. Few of those commenting on the genome project in these studies are laboratory molecular biologists familiar with polymerase chain reaction, in situ hybridization, or any of the other technologies for manipulating DNA that have been so important to the project. They are,

instead, scientific outsiders who are expected to shed light on the long-term social implications of the access to hereditary information that the genome project promises to make possible.

Justice and the Human Genome Project is a collection of relevant essays by scholars from many disciplines. Insurance executive Robert J. Pokorski, for example, presents an overview of the fundamental principles of private health insurance, and in so doing makes clear the institutional forces that will shape the uses of genetic information. He constructs his essay as a defense of the insurance industry and its underwriting policies. Those policies are intended to limit risk and thereby, as he puts it, protect those who are healthy. The "great majority of applicants whose genetic information is favorable" should not be "forced to pay higher rates so that those at greater risk could pay less than is required by an equitable estimate of their own risks." He forthrightly states that the health insurers will use predictive genetic information to exclude some people or to charge them higher rates, but, he says, such a plan is not discriminatory because it is based on the subscriber's actual health status. Insurance policies are only discriminatory, according to Pokorski, if they are not based on "sound actuarial data." He says that insurers "fear most" that applicants will withhold information about their genetic risks, thus gaining "an unfair advantage" in the application process. He further suggests that Americans "choose the type of insurance system they want" and, since they have chosen private insurance, they have likewise chosen restricted access to medical care for those who can be expected to be a financial burden to the system.

An endnote proclaims that the opinions expressed in the essay are those of the author, not of the insurance industry. The actuarial logic that Pokorski carries to its conclusion is, nonetheless, the guiding ideology of that industry, and Pokorski has dramatically captured the rationale for denying insurance to people who are found to be at risk of genetic disease. It is perhaps ironic that this rationale should be so well-explained in a book with the word "justice" in the title. I think Pokorski's chilling essay should be required reading for anyone promoting the quaint idea that acquiring genetic information is an unmitigated good for the victims of genetic disease.

Informative in a different way is legal scholar Lori Andrews's essay on the legal regulation of genetic testing. Andrews has categorized the types of legislation that either allow, forbid, or require the use of genetic testing. As she explains, the United States has already seen two waves of laws devoted to forcing people to use genetic information. The first wave occurred at the height of the American eugenics movement during the first three decades of the twentieth century, when many state legislatures mandated involuntary sterilization of those deemed unfit (and therefore a costly burden to the state). The second group of laws came in the 1970s and focused on mandated genetic screening of African-Americans for sickle-cell trait, this time with the intention that such information could help carriers make reproductive decisions. Both mandatory screening programs were disastrous for those affected. As a consequence of eugenics laws, many were sterilized on the basis of limited information—a social worker's assessment that they were "feeble-minded," for example. In the sickle-cell screening program, those found to carry the trait

faced job discrimination, psychological trauma, and even social ostracism, partly because little counseling or community education was available to explain the tests and their meaning.

Mandatory genetic screening does persist in newborn screening programs for phenylketonuria (PKU) and (in some states) other metabolic disorders. PKU is a hereditary metabolic disorder that causes mental retardation unless a restricted diet containing no phenylalanine is adopted at birth and maintained for many years (some claim five years is long enough; others that a lifetime is necessary). Mandatory newborn screening for PKU was widely adopted in many states after a simple blood test for detecting the disorder was developed in 1961, and PKU is commonly held up as an example of the dramatic potential of genetic testing to help both individuals and society. But Andrews argues that PKU testing could be just as effective if it were voluntary. She is not interested in eliminating PKU testing but is concerned about the implications of state-mandated genetic tests, and she sees PKU as the beginning of testing programs that could easily expand. Andrews is implicitly interested in the possibility of a "new eugenics" driven by medical practice and legal pressures. Other essayists make this interest more overt.

Particularly relevant here is historian Daniel Kevles's essay on eugenics and the Human Genome Project. Eugenics is of course a slippery concept that can be defined in many ways, and Kevles here defines it first very narrowly, as state-mandated control of reproduction by forced sterilization or by the establishment of government incentives (cash, for example) to encourage the more genetically fit to reproduce. This sort of eugenics, he predicts, is unlikely in the United States. First, he suggests, gene mapping is not likely to produce reliable information about the qualities the world most admires—"talent, behavior, personality." Furthermore, the technology required to produce "designer genomes" is neither available now nor, he believes, likely to be available any time in the near future. These conditions are, in Kevles's formulation, the technical barriers to a new eugenics, but there are also social barriers. These include an "awareness of the barbarities and cruelties of state-sponsored eugenics in the past" that has "tended to set most geneticists and the public at large against such programs."

After defining eugenics narrowly and proclaiming it unlikely to return for both technical and social reasons, Kevles then states that contemporary medical genetics poses "real social challenges," and these include the possibility of a "homemade eugenics" (in Robert Wright's phrase) mediated by parental love. Every parent wants a healthy baby, and as it becomes possible for parents to "choose the kinds of kids they want to have," they can be expected to choose not only children who are healthier but also more intelligent, or better looking, or more athletic. Kevles also recognizes that genetic diseases are costly—to the state and the health-care delivery system—and any future system of universal national health care could, under the pressures of rising costs, eventually compel people not to bear children with genetic disease, "not for the sake of the gene pool but in the interest of keeping public health costs down."

A much more fanciful set of predictions appears in bioethicist Arthur Caplan's exploration of the dilemmas posed by genetic information. I must admit to a certain bias, as a historian, against hypothetical case studies, but Caplan's six imaginary stories about

genetic testing in 2030 are both entertaining and provocative. In one, a Minnesota Jew who can "no longer stand the sterile, artificially controlled climate of his home state" seeks to emigrate to Israel. To do so, he must pass a genetic test that will determine whether he is of proper Jewish matrilineal descent. But the man's maternal grandmother was not Jewish. Is it fair, Caplan asks, for genetic testing to be used to identify who is and is not a "real" or "true" member of a racial group? In another scenario, a Native American tribal leader, seeking to qualify for a special scholarship program, is discovered to have too many genetic markers in common with the "white" population. She is one of the few remaining native speakers of her tribal language and is active in her tribe's affairs, but she is also apparently "white." "Should social policy allow people to define themselves on the basis of culture and behavior as belonging to a particular group, or is biological inheritance a key element of membership as well?" In still another scenario, a researcher accidentally discovers that Jimmy Carter, whose DNA is being used in a research program, has a marker that indicates he had an African-American forebear. Should this information be made public? How should unintentionally acquired information about race or ethnicity be handled?

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Caplan demonstrates that information about biologic differences in races already influences the distribution and allocation of medical resources. In some organ transplantations, matching antigens greatly improve success rates. Such antigens differ in different racial groups, with the result that organs from white donors almost always go to white recipients. In the case of kidney transplantation, this means that blacks have a much lower likelihood of receiving a donor kidney, for African-Americans are both more prone to kidney disease and less likely to donate their organs. As Caplan indicates, the ethical problem of race classification is "one of the greatest moral challenges" posed by modern genetics.

Finally, legal scholar George Annas proposes some rules for gene banks that emphasize the private nature of genetic information. Ideas about what counts as private information have shifted dramatically in recent years. Biographers have gained access to tapes of psychotherapy sessions (for example with Anne Sexton's therapist) and journalists to medical information about celebrities (Earvin "Magic" Johnson's physician, for example, released private medical information to a *New York Times* reporter after Johnson announced that he was infected with HIV). Meanwhile presidential candidates

now routinely expect to release medical information to the public. By exploring both existing practices in medical record-keeping and the use of DNA in criminal records, Annas suggests that current standards are inadequate. He proposes that gene banks in the future should follow strict privacy guidelines, which he sketches out in the essay. These include criminal penalties for the misuse or unauthorized use of genetic information and tightly controlled access to DNA samples. Although "it may seem premature to develop rules or guidelines for DNA banks," Annas comments, we are fast approaching a time when such rules will be necessary. And although most of us will not be the focus of biographies or police investigations, we are all likely to be affected by the interests of insurers, employers, and families who have a stake in the predictive information contained in our DNA.

Philosopher Timothy Murphy approaches some rather different questions about the project when he asks whether it should be undertaken at all. As he notes, some commentators have suggested that the scientific work itself poses no moral dilemmas. It is only when the data are used that they come to have ethical significance. Murphy interprets this view as reflecting shared assumptions about the value of scientific research rather than empirical truths about the value neutrality of the scientific enterprise. "Is it true that there is no moral substance in the genome project itself?" he asks, then answers his own question by laying out the moral questions the scientific work raises. These include questions about priorities, about who will benefit from the research, and whether genomics simply diverts attention from social solutions to problems.

Also included are an essay by philosopher Leonard Fleck, who places genetic technologies in the context of other medical technologies for preserving health, and one by philosopher Norman Daniels on the problems genetic information poses for a health-care delivery system. Marc Lappé closes the volume with an examination of ideas about difference, equality, and their relationship to genetic testing.

The essays in *Genes and Human Self-Knowledge* represent a much wider range of perspectives and a correspondingly wider range of quality. These essays grew out of papers presented at a 1992 conference at the University of Iowa; the finished text contains papers and short commentaries on some of them. Contributors include historians Mitchell Ash, Susan Lawrence, and William Carroll, historian and political scientist Diane Paul, philosophers Evan Fales, Dan Brock, Panayot Butchvarov, David L. Hull, Michael Ruse, and David Magnus, biologist Ruth Hubbard, medical geneticist Kimberly Quaid, health-science journalist Larry Thompson, and high-school science teacher Kevin Koepnick. As this range suggests, the conference was intended to bring many voices into the debate.

Diversity is one of the reigning values in the contemporary academy, and it has its merits; in this case, however, it has resulted in a volume held together solely by the binding material. The theme of "genes and human self-knowledge" is sufficiently broad to encompass essays on free will, reductionism, and other philosophical topics, as well as on the history of eugenics, the experiences of persons with genetic disease, and the ethics of human germ-line intervention. Some of the essays included are interesting and

well conceived, despite the book's lack of cohesion. I want particularly to mention Diane Paul's sensible discussion of eugenics, a discussion that (again) considers the problem of defining this fuzzy concept. "One can be opposed to eugenics, and for almost anything," Paul states. Defining eugenics as limited to state coercion would violate history: "Francis Galton would no longer be a eugenicist. Nor would H. J. Muller or William Schockley." She suggests that it is more productive to try to understand what critics fear when they state that genomics will lead to a new eugenics.

Interesting from a different perspective is Kimberly Quaid's essay on victims of Huntington disease. Huntington disease is a late-onset dominant genetic disorder that causes personality and mood changes, depression, dementia, and disturbances in both voluntary and involuntary movement. The average age of onset is 38; those affected live an average of 16 years after diagnosis, but much of that time is spent in a nursing home in almost complete dependence. Quaid is a "wise woman," one who is "normal," but whose special situation has made her "intimately privy to the secret life of the stigmatized individual and sympathetic with it." She has counseled those seeking presymptomatic testing for Huntington disease for five years, and this has permitted her to understand what the genetic test, and the disease, have meant to families and to individual patients. The stories collected here are gripping, as patients describe and defend their choices (to be tested or not, to have children or not, to give up on life or not). Quaid does not attempt an analysis of these comments, or of the status of those who test positive. She is interested in the people and their experiences. Her essay is a rare opportunity to consider the impact of genetic testing from the perspective of those tested.

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The remaining essays include an informative description by LeRoy Walters of the technical possibilities raised by the Human Genome Project, a fairly substantive discussion of genetic discrimination by Larry Gostin, and two essays on the public understanding of science by Joseph D. McInerney and Larry Thompson.

Let me close with a brief description of Tom Wilkie's *Perilous Knowledge.* Wilkie is a former physicist turned science editor and journalist. His book is basically a broad overview of the issues raised by the new genetics. Although his subtitle identifies his subject as the Human Genome Project, the text actually deals with a wide range of genetic technologies and ethical dilemmas that predate the genome project and are not in any way specific to it. The book traces the history of gene therapy, human gene mapping since the 1960s, the technologic innovations that made possible direct manipulation of human

DNA, and the institutionalization of the genome project in the 1980s and beyond. His final chapters explore cases in which the new ability to identify and manipulate human genes has raised ethical questions: in testing for sickle-cell anemia, in the use of human growth hormone for children who are interpreted as too short by their parents, and in gene therapy in general. The author has an engaging writing style and the ability to explain scientific terms and ideas in accessible ways. I cannot, however, recommend the book as a general introduction to contemporary human genetics and its ethical implications because it includes no footnotes, references, or bibliographic material of any kind. Wilkie has obviously drawn on the large and growing literature on medical genetics, including the many participant histories that have appeared in the last few years, but he has made it impossible for those interested in the topics he explores to pursue them further. This limits the usefulness of his text.

In all, these volumes are not very encouraging. Some scholars seem willing to comment on the genome project and its implications without having done much research on the subject. But the institutional and social implications of the genome project are not simply matters of opinion resolvable through hypothetic musings. Understanding them depends on a knowledge of medical and legal practice, of the organization of the scientific community, and of the historical development of human genetics.