

# Chapter 19

## The Human Genome Project

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*A more important set of instruction books will never be found by human beings. When finally interpreted, the genetic messages encoded within our DNA molecules will provide the ultimate answers to the chemical underpinnings of human existence. They will not only help us understand how we function as healthy human beings but will also explain, at the chemical level, the role of genetic factors in a multitude of diseases—such as cancer, Alzheimer's disease and schizophrenia—that diminish the individual lives of so many millions of people.*

James Watson<sup>1</sup>

Medicine in the twenty-first century will be dominated by genetics. The Human Genome Project, the multi-billion-dollar international attempt to map the human genome, which was born of a powerful alliance between governments, private enterprise, and science, has been described as a Holy Grail or Rosetta Stone for deciphering the secrets of humanity and all biology that, until now, nature has adeptly guarded and kept just beyond science's grasp. The working draft human DNA sequence has revealed that within the genome's 3 billion bits of information, there are in the range of 35,000 human genes, far fewer than the expected 100,000, and only a little more than twice that of a fruit fly, a mosquito, or a worm. Still, the larger volume of data associated with the larger human genome makes for a considerable increase in complexity. The human genome (the first vertebrate genome) is around 30 times larger than those of the fly, worm, and mosquito, and 250 times larger than the yeast (the first sequenced eukaryotic genome).<sup>2</sup> The genomes of more than 180 organisms have been sequenced since 1995.<sup>3</sup> In 2002, scientists from 27 institutions in six countries reported the decoding of the complete set of genes of a mouse.<sup>4</sup> The mouse code consists of 2.5 billion base pairs (only slightly smaller than that of a human), 5% of which are identical to humans—perhaps lending some credence to scientists' theory that mice and humans shared a mammalian ancestor.

"What a piece of work," as Hamlet marveled, "is a man."<sup>5</sup> While some view the early successes of the Human Genome Project as a magnificent and powerful basis of discovery that will profoundly alter the way we view ourselves, some fear the Human Genome Project is an attempt to reduce man, that marvelous "piece of work," to biology, which could lead us back to the road of the eugenics of old. Still others

warn of a growing unease in the public sphere of a massive control of the processes of life, its potential social dangers, and a "fear of Frankenstein."<sup>6</sup> For every enthusiastic claim of its value, there has been an apocalyptic counterclaim. Notwithstanding what will certainly be an ongoing controversy and debate in scientific, political, theological, and lay circles over the merits of genomics and the role of genes in human life, our understanding of how genes function continues to expand at an almost unfathomable pace. As genetic technology relevant to human biology is developed, society will be challenged by a host of complex ethical, legal, and social issues. Although many of the technologies discussed here and elsewhere related to the Human Genome Project are not yet available, it is not too soon to begin a dialogue and a careful analysis of the potential costs and benefits of future genetic programs and to formulate a tentative plan for how society and medicine might respond.

### HISTORY OF GENETICS

Gregor Johann Mendel, an inquisitive Augustinian monk and priest who had studied at university in Olmütz and Vienna, had a passion for tending his pea plants in the gardens of the monastery in Brnn, Austria (now Brno, Czech Republic).<sup>7</sup> In 1856, his fascination with the distinct varieties of plants and the resulting peas led him to begin an 8-year series of experiments that would involve the planting of over 30,000 different plants, the crossing of his pea varieties, and recording the types of plants produced. What may have seemed an obsessive interest in gardening led to his discovery of the elements of heredity, which he called genes, and some basic rules governing the transmission of traits from generation to generation. In 1865, he presented the results of his work to the Brnn Society for the Study of Natural History and those results were published in 1866 in *Experiments with Plant Hybrids*.<sup>8</sup> Though his work revolutionized biology and experimental science, its importance was not realized in his lifetime. Not until 1900 was his work rediscovered and it was later still that he would be hailed as the father of modern genetics.

In 1859, the first edition of Charles Darwin's *On the Origin of Species* was published and sold out in one day. In it he described the modern theory of evolution: adaptation

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is the result of natural selection. Natural selection is a consequence of hereditary differences among organisms in their ability to survive and reproduce in the prevailing environment.<sup>9</sup> Those best suited to their environment will have better survival and reproductive success than others of their kind and will then pass those advantageous traits to their offspring. Although Darwin could provide a detailed account of how the mechanism of natural selection worked, he was unable to explain how variations could arise in populations or how those characteristics could be transmitted from one generation to the next.<sup>10</sup> Although Darwin and Mendel were contemporaries, neither was apparently aware of the other's work. By 1900, long after the death of both Mendel and Darwin, three botanists working in three different places on different species, duplicated Mendel's work before his original paper was rediscovered. In 1918, in Britain, the mathematical genius of Ronald Fisher reconciled Darwinism and Mendelism, proving that Mendel had brilliantly "supplied the missing parts of the structure erected by Darwin."<sup>11</sup>

In 1951, chance threw a precocious, confident 19-year-old American, James Dewey Watson, with a bachelor's degree from Indiana University, into the Cavendish Laboratory at Cambridge University with 35-year-old Francis Harry Compton Crick, a British biophysicist (who had yet to attain a PhD). Both were convinced that genes were made of DNA, not protein—as was commonly believed at the time. Together they began an inquiry into the structure of DNA. Utilizing x-rays and molecular models, by 1953 they proposed the first essentially correct three-dimensional representation of the double helix structure of DNA and could explain how the DNA molecule replicates, controls heredity, and undergoes mutation. Their discovery paved the way for all the major genetic discoveries of the last 50 years. In 1962, Watson and Crick were awarded the Nobel Prize for Medicine, for their "discoveries concerning the molecular structure of nucleic acids and its significance for information transfer in living material," a prize they shared with Maurice Hugh Frederick Wilkins, who simultaneously worked on the structure of DNA along with Rosalind Franklin (deceased and therefore unable to be honored with the Nobel Prize) at King's College in London. Watson and Crick have since been variously characterized as having made the greatest scientific discovery of the twentieth century and one of the greatest discoveries *ever* made.

## HISTORY OF THE HUMAN GENOME PROJECT

The Human Genome Project (HGP) was a 15-year effort formally begun in October 1990. At that time, its stated goal was to analyze the structure of human DNA, determine the location of all human genes, and make them available for further biological study, with parallel studies being carried out on selected model organisms to provide the comparative information required for understanding the functioning of the human genome. Another long-range project goal is to determine the complete sequence of the

3 billion DNA base pairs in the human genome, a feat that even with today's technology remains awesome to consider.

The groundwork for the project began in 1985 when Charles DeLisi, Director of the Office of Health and Environmental Research (OHER) at the Department of Energy (DOE), the division responsible for funding most of the life sciences and environmental research for the department, proposed a Human Genome Initiative. His interest in the project grew out of an effort to study DNA changes in the cells of atomic bomb survivors of Hiroshima and Nagasaki, and to augment the ongoing work of the Atomic Bomb Casualty Commission in its study of the biological effects of radiation exposure.<sup>12</sup> The DOE also had an interest in being able to utilize high technology national laboratories and their multidisciplinary teams of scientists.<sup>13</sup>

Soon after the initial proposal, the OHER convened a conference in Santa Fe, New Mexico, to assess the feasibility of a Human Genome Initiative. Following the Santa Fe conference, DOE's OHER announced the Human Genome Initiative. A year later, in 1987, the Congressionally chartered DOE advisory committee, the Health and Environmental Research Advisory Committee (HERAC), endorsed the plan for a 15-year, multidisciplinary scientific and technological undertaking to map and sequence the human genome. In 1990, the DOE and the National Institutes of Health presented a joint HGP plan to Congress and the 15-year program formally began. Also in 1990, recognizing the wider social implications of the HGP, the Ethical, Legal and Social Implications (ELSI) Program was established as part of the HGP to identify problem areas and develop solutions before newly gained scientific information would be integrated into health care practice.<sup>14</sup>

The progression from 1985 to 1990 was not an easy one for the DOE. It would have to explain why the agency should play a major role in the project and explain its connection and expertise relative to the project.<sup>15</sup> Further, as the proposal for the HGP was thrust onto the public policy agenda, the project's goals had to undergo several "redefinitions" to ensure that each hurdle, whether it be in the Senate or the House or one of many hearings, would be cleared. At least part of the redefinition was aimed at seizing the lay imagination with the excitement of the possibilities of more robust health and cures for disease.

In 1992, amid differing scientific aims and conflicts about his patent rights, NIH scientist Craig Venter resigned his post to head up a new Institute for Genomic Research with hopes of speeding up the project using state-of-the-art automation and avoiding the bureaucracy of operating in the public arena. By 1994, the project had announced that its 5-year goal of genetic mapping was one year ahead of schedule. That same year, the Genetic Privacy Act was written—the first United States HGP legislative product, proposed to regulate collection, analysis, storage, and use of DNA samples and genetic information obtained from them.<sup>16</sup> Still farther ahead of schedule, on June 25, 2000, the HGP leaders, both public and private, and President Clinton announced the completion, 5 years early, of a "working draft" DNA sequence of the human genome.<sup>17</sup> That initial working draft sequence was published on

February 12, 2001.<sup>18</sup> Finally, on April 14, 2003, the International Genome Consortium, led in the United States by the National Human Genome Research Institute (NHGRI) and the DOE, announced the successful completion of the HGP more than 2 years ahead of schedule.

## THE NEW GENETICS AND THE SHADOW OF EUGENICS

George Santayana, a U.S. philosopher, is credited with penning the dictum, “[w]hen experience is not retained ... infancy is perpetual. Those who cannot remember the past are condemned to repeat it.” Eugenics was the early twentieth-century movement for hereditary improvement that was wrought with virulent racism and atrocious wrongs visited upon vulnerable individuals and populations. Some, remembering the past, fear that mapping the human genome opens up the possibility of widespread nonmedical uses of genetic knowledge. Eugenics and its attendant social and moral failures, promoted by a particular conception of human perfection, led genetics into the bleakest period of its short history.

Although Francis Galton (1822–1911), a cousin of Darwin and a respected and influential British scientist, is given the dubious honor of being the creator of eugenics,<sup>19</sup> the eugenic ideal has been a part of Western discourse at least since Plato, who proposed, in Book Five of the *Republic* (459d), the selective breeding of the guardians to ensure superior offspring (positive eugenics), coupled with the destruction of those deemed inferior (negative eugenics).<sup>20</sup> Plato, however, never garnered the momentum that the eugenic movement achieved on both sides of the Atlantic before World War II. With eminent geneticists poised at the forefront, popular eugenics was rapidly introduced into the public discourse.<sup>21</sup> Further, eugenicists drawing on Darwinian notions of evolution viewed medical care as a frustration to evolution by allowing the unfit to survive and reproduce.<sup>22</sup>

Eugenics came to be viewed as a completely respectable part of American and British biology, taught at universities and incorporated into laws.<sup>23</sup> In the United States, by 1911, six states already had laws allowing forced sterilization of the mentally unfit. By 1917, nine more states had joined them. The Immigration Restriction Act of 1924 was a direct result of eugenic campaigning.<sup>24</sup> That law remained unamended for 40 years. In 1927, Justice Oliver Wendell Holmes of the United States Supreme Court, in his majority opinion in *Buck v. Bell*, ruled that the Commonwealth of Virginia could sterilize Carrie Buck, a 17-year-old girl, deemed feeble-minded in light of eugenic theory.<sup>25</sup> Similar sterilizations continued in Virginia until 1972—four were performed that year. The laws allowing those sterilizations are still on the books in Virginia, and similar laws remain in effect in many other states.<sup>26</sup> The Supreme Court has never reversed its decision in *Buck v. Bell*. The United States, where individual liberty is so highly valued, crudely interfered with the rights of individuals to determine their reproductive futures by sterilizing more than 60,000 people for “feeble-mindedness” between 1910 and 1935.<sup>27</sup>

Eugenics brings to mind a time when “science” served to justify social prejudice by its obsession with biological improvement of the human race. The evils transpired when the values of a few were mistaken for the values and goals of human beings in general. Though no one may expect a repeat of the grizzly racial policies of Nazism, history does account for a level of mistrust of genetic science. There is a sense that an ethically ambiguous and potentially destructive body of research is taking place, conducted by scientific elites, that promises to alter our lives in some dramatic or undesirable way.<sup>28</sup> Some critics fear that a subtle form of eugenics may slip in through the cultural backdoor.<sup>29</sup> The growing technological abilities to control human genetic makeup could foster the emergence of the image of the “perfect child,” and the impact of the social value of perfection will begin to stigmatize and oppress all those who fall short. Knowledge of the past is an indispensable guide to the future. It is not difficult to understand the fear that a benign policy of forestalling disease may become a program for enforcing social prejudice and a new eugenics. Even taking care to remember the past, some question whether society is well enough aware of the problem to avoid it.

## GENETIC REDUCTIONISM/ DETERMINISM

*We cannot allow any barrier to stand in the path of our complete control and thereby understanding of the life phenomena. I believe that anyone will reach the same view who considers the control of natural phenomena is the essential problem of scientific research.*

Jacques Loeb<sup>30</sup>

This sentiment, noted among scientists to some degree since Descartes,<sup>31</sup> has grown into what some refer to as a Reductionist Revolution in biology, genetics, and the life sciences. As the HGP moves forward toward its goal of identifying each base pair among the 3 billion that comprise the human genome, there is concern that scientists will become more persuaded that genes alone are the control centers, not only of whether we are tall or short, have green eyes or brown, or have a predisposition to develop heart disease, but also whether we behave in certain ways or entertain certain thoughts.<sup>32</sup> Critics argue that humans will be reduced to the sum of their genes—at the formation of the zygote, the future potential of the person it will become will be known or knowable—genes will be equated with destiny, and all will be consigned to the “Trap of Determinism”—condemned to a fate written in their genes before they were born.<sup>33</sup>

Reductionism hearkens back to the Hobbesian notion that human beings are “merely complex machines.”<sup>34</sup> It presupposes that all of the phenomena of biology, even that of human nature, obey the laws of chemistry and physics. The previously unimaginable successes of molecular biology realized in the HGP and in our understanding of the machinery of the cell have given the impression that

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no problem is beyond the analytical power of science—that even the genome is concrete and manageable so that complete knowledge of the entire organism is just around the corner.<sup>35</sup>

Another criticism of the growing trend toward reductionism is the concern that a focus on the “power” of the genes will lead to a lessening of efforts to improve public health by correcting environmental contributors to disease.<sup>36</sup> “The most serious objection to predisposition studies... is that they can detract attention from the epidemiological fact that cancer is a disease whose incidence varies according to occupation, diet, socioeconomic status, and personal habits such as smoking.” Ignoring such epidemiological relationships would be dangerous in terms of setting public policy. Antipollution laws or laws governing workplace safety might become low priority if it is individuals’ genetic composition that really provides the risk.

### GENETIC ENHANCEMENT

*Health is a state of complete physical, mental and social well-being and not merely the absence of disease or infirmity.*<sup>37</sup>

As knowledge and capabilities in genomics mature, there is little controversy that efforts, for the most part, should be directed toward interventions to prevent or cure disease.<sup>38</sup> If health is more than the absence of disease, however, genomics could offer a future that redefines physical, mental, and social well-being. Those new definitions may “ratchet up” the standard for “normal” human functioning, and disability and insurability may likewise be redefined. A difficult question that society will have to consider is whether “genetic enhancement”—that is, use of genomic technology to select height, eye color, temperament, or intelligence of future offspring—is morally permissible.<sup>39</sup> Genetic enhancement involves no abnormal gene; its goal is to amplify “normal” genes to make them “better.” Genetic science has a long way to go before enhancements are developed, are proven safe and effective, and become widely available commercially. Notwithstanding, as the technology required for such genetic enhancements move closer to being realized, should we celebrate the fruits of biotechnology’s labors that make it all possible, or should there be limits and controls on their activities and society’s choices? The profit motive and the inexorable law of supply and demand are among the strong social forces that will make it extremely unlikely that genetic technology will be limited to preventing and curing disease.<sup>40</sup>

When considering the possibility of making genetic enhancements available, a critical first step will be to ascertain *which* enhancements are best for future children and *who* will decide what kinds of children would be best.<sup>41</sup> The history of the eugenics movement and the frequency of racist attitudes about what characteristics would be desirable should provide reasons for pause.<sup>42</sup> In answering the “who” question, the right to privacy would necessarily extend to enhancement decisions—recognizing the importance, for parents and children, of parents having substantial discretion

and freedom to decide how to raise their children.<sup>43</sup> Parents may be just as susceptible as the early proponents of eugenics to such stereotypes and prejudices.<sup>44</sup> At the time when an enhancement decision is being made, it may be difficult to separate out individual “parental” choice from the larger societal influences at play. What may be wrong with eugenics is that the social currents coerce in ways that we scarcely notice—our individual “choices” becoming mere instantiations of prevailing cultural imperatives about what bodies, gender, and capacities of intellect are desirable.<sup>45</sup> Although history provides strong grounds for caution about attempts to perfect our children, if genetic interventions that assure future generations will be beneficiaries of genes that will enable their lives to be better can be pursued justly, they should not be abandoned.<sup>46</sup> Moreover, new molecular knowledge must be evaluated by considering the social surround at the time an enhancement is considered, not with the level of knowledge and understanding we have today. In spite of the past, or perhaps because of it, the moral permissibility of eugenic goals must be addressed on its own terms.

### ACCESS TO GENETIC SERVICES

*...We hold these truths to be self-evident: that all men are created equal; that they are endowed by their creator with certain inalienable rights; that among these rights are life, liberty and the pursuit of happiness.*

*Declaration of Independence*

Our domestic political history has been dominated by the demand for equality and the resistance to that demand.<sup>47</sup> In a world that tolerates so much inequity in the circumstances into which its children come into being, it is hard to make the argument that there lies a moral distinction between their rights to biological advantages as opposed to social and economic advantages.<sup>48</sup> Because of the urgent social problems present long before the HGP was proposed, the beautiful world that its enthusiasts envisage, of a kind of “genetic utopia,” may be a long way from reality.<sup>49</sup> Already we struggle with the fair allocation of vital health care resources in a society marked by great discrepancies in wealth and social status.<sup>50</sup> The likelihood that genetic technologies will be available to some people but not to others, and that a major determinant of access will be wealth, raises profound social issues.<sup>51</sup> Lack of insurance coverage combines with other factors of the delivery system to have a systematic effect on the utilization of health care services.<sup>52</sup>

Wealth-based access to health care has always been a distinct feature of our health care system. Those patients able to afford the service can simply “buy” it, even if it is not covered by a private or public insurance plan. The failure of most insurers to pay for in vitro fertilization will narrow the pool of patients able to access some genetic services to the most wealthy.<sup>53</sup> Likewise, if insurers define genetic enhancements as “not medically necessary,” as they do cosmetic surgery, then only those able to purchase them with personal funds will be able to access them; the

more expensive the enhancement, the more limited this group may be.<sup>54</sup>

Aside from economic barriers, the benefits of genetic information are likely to be available only to some people: those who know about genetic testing, who know how and where to get it, and who can assimilate the results.<sup>55</sup> Those who are able to access the new technologies will do so as a definitive preventive measure, as a therapy for many disorders, or they will embrace genetic enhancements as a means of gaining socioeconomic advantages. In the worse case scenario, this unequal access could conceivably lead to a “genetic aristocracy” or “genobility”—those with genetically engineered advantages that enable them to monopolize the most lucrative jobs and investment opportunities, thus widening the gulf between them and the “genetic underclass.”<sup>56</sup> If, as some argue, the HGP has been oversold, it may be because things have been made to appear simpler than they are.

Norman Daniels, a noted American philosopher, posits that there is something special about health care—something distinguishing it from other social goods such as food or clothing. He suggests that if there is a right to health care, it is because of the kinds of needs it meets.<sup>57</sup> A right then presupposes an obligation on the part of the state to make health care services available on the basis of medical need, without regard to ability to pay or other nonmedical factors.<sup>58</sup> The “specialness” of health care requires that it be treated differently from other social goods, even in a society that tolerates (even glorifies) significant and pervasive inequalities in the distribution of most social goods.<sup>59</sup> Many agree that it is a requirement of justice for health care to be distributed more equally.<sup>60</sup>

If there is indeed a “right” to health care, is it identical to a right to receive genetic therapy? Assuming some scarcity of resources and the requirement that health needs be met under reasonable resource constraints, would genetic “disease” be a social need that the state would be obliged to insure against, or would it be responsible to provide access only to a basic minimum? Is some level of health care and “genetic care” rationing inevitable? Daniel’s theory of *Just Health Care* puts forth a hierarchy of *needs*, suggesting that there may not be an obligation, even in a theoretical world, on the part of the state to provide access to all available services.<sup>61</sup> He maintains that individuals must have a fairly equal opportunity to obtain those health care services that will provide them a “normal range of opportunity.” That range of “normality” may change at the same pace as new developments in genetics.<sup>62</sup>

## GENETICS AND PRIVACY: INSURANCE AND EMPLOYMENT

Given the rapid progress of biotechnology and bioinformatics in recent years, coupled with the successes of the HGP, the volume of available genetic information is on the verge of an explosion.<sup>63</sup> Because proliferation of such information could result in social stigma and the loss of educational and social opportunities and highly valued

freedoms, societies with strong commitments to autonomy and individual rights will be challenged to develop the best strategies for protecting privacy.<sup>64</sup> Scientific, technological, and cultural developments alter the context in which the need for privacy exists. These advances push societies onto uncharted ethical and legal terrains. There is no property so private, no information more confidential, than a person’s genetic profile. Therefore, the premium to be placed on an individual’s rights is greater when the issue is something as personal and as intimate as their genetic constitution. In the ever-changing, ever-advancing milieu of genetic science, society must adapt and extend privacy protection to conform to the environment.

Inherent features of genetic information make it qualitatively different from other forms of health data. It relates to a range of people, not just one individual. Information revealed about the individual who consented (the proband) to the genetic testing may also reveal personal information about the proband’s parents, siblings, and children, and may have implications for the spouses or potential spouses (and future offspring). This gives rise to special concerns for how the information is gathered, stored, accessed, and used. Another feature that distinguishes genetic data from other forms of health information is that it is not merely about one’s medical past, but can also furnish information about one’s medical future (and that of blood relatives). Because of these unique characteristics of genetic information, many believe that the concern for privacy should be enhanced. Others believe, however, that there should not be a strong unflinching commitment to privacy protection at the expense of other social goods and that individual liberty must yield to the greater needs of the community.<sup>65</sup>

What kinds of harms are threatened by the loss of genetic privacy? At worst, there is a risk that knowledge generated by the HGP, if freely disseminated, could lead to the creation of a subcaste of “genetic lepers” who are refused jobs, insurance coverage, and even possibly the right to marry and have children.<sup>66</sup> Today, a primary concern is that insurers will use information regarding an individual’s propensity to develop an illness as a preexisting illness to justify denying, limiting, or canceling insurance policies, or, alternatively, to charge prohibitively high premiums. Another concern is that employers will use genetic information against current workers or to screen potential employees and then use that information to refuse to hire (or as a reason to fire) individuals with genetic predisposition to disease to avoid having to pay for or to supplement the costs of future care.

An individual’s autonomy and privacy rights as concerns genetic information mean the right to control what they or others may know about them. The highest level of control may be exerted through controlling the initial production of information. Although the HGP has inspired enthusiastic and hyperbolic descriptions from supporters, when risks of social stigma and loss of insurability are high, and when no medical intervention exists for positive results, one may reasonably decide that the risk/benefit ratio is too high and refuse testing.<sup>67</sup> Protective measures may need to

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be implemented, however, to assure that those who refuse testing do not suffer repercussions that reverse that risk/benefit ratio. For example, if an employer can refuse to hire based on a prospective employee's refusal to submit to genetic tests, any right to privacy would protect only if being unemployed is a tenable option.

Control of disclosure of genetic information by determining the identity of recipient(s), the purposes for which the information is used, and the period of time within which future disclosures may be made, is often lost after the initial consent to disclose is given. For example, stored Newborn Screening Guthrie cards could become the basis for establishing DNA databases for new and previously unimagined purposes if their use is not regulated.<sup>68</sup> Protections will have to be tailored to account for the unusual predictive power of genetic tests that makes their results useful to third parties whose interests may not always be innocuous.

To date, there has been no federal legislation directed to the novel challenges to privacy that the HGP and associated new technologies will present, although several bills were introduced during the last decade.<sup>69</sup> However, on February 8, 2000, President Clinton signed an executive order that prevents every federal department and agency from considering genetic information in any hiring or promotion actions, thereby providing some protections for federal employees. Nearly every state has enacted some form of genetic nondiscrimination laws, none of which is comprehensive.<sup>70</sup> The most likely source of protection against genetic discrimination in the workplace is Title I of the Americans with Disabilities Act of 1990 (ADA), enforced by the Equal Employment Opportunity Commission (EEOC).<sup>71</sup>

The Health Insurance Portability and Accountability Act (HIPAA) provides some protections against genetic discrimination in the realm of insurance, but it applies only to employer-based and commercially issued group health insurance. There is no similar law that protects private individuals seeking health insurance. HIPAA does, however, provide protection for information in a patient's medical record, regardless of their insurance status. The new federal standards limit the nonconsensual use and release of private health information (not specific to genetics) and generally restrict any release to the minimum needed for the intended purpose.

Existing legislation is a move in the right direction, but comprehensive privacy protections will become more urgent as the economic incentive to discriminate based on genetic information increases and the costs associated with genetic testing decrease. Insurance companies are always seeking ways of identifying high risk groups. The insurance industry could rightly claim that stringent privacy legislation would place them at an unfair disadvantage. Those less than optimistic about the efficacy of privacy laws to prevent unauthorized dissemination of genetic information believe that the focus should be on implementing antidiscrimination laws that would proscribe the *use* of genetic information should privacy safeguards fail.

## CONCLUSION

*Now is not the end. It is not even the beginning of the end.  
But it is, perhaps, the end of the beginning.*

*Winston Churchill, 1942, after 3 years of war*

As the Human Genome Project (HGP) continues toward its goal to identify each of the 3 billion base pairs that make up the human genome, some believe the solution to the mysteries of humanity, the most magnificent and complex biological Rosetta Stone, may be near. The seemingly disparate paths of the sciences of molecular biology, chemistry, and physics have converged to this end. The work of the HGP and the knowledge it spawns will someday reveal a new human anatomy. As James Watson has recognized, "[i]f society is to cope with the consequences of this knowledge, people must learn and become better informed about genetics."

Proponents of the HGP view it as a trustworthy and badly needed step toward improving the health of mankind, but ethical debates must acknowledge the horrors perpetrated in the name of eugenics in the past century. The important dialogue already begun, must continue with a focus on forestalling the threat of creating new forms of discrimination and methods of oppression. Much of the suspicion of the HGP is a presumed hidden agenda to control the future of humanity through manipulating human genes and the reductionist philosophy that humans are "determined" by their genes.

Almost everyone would agree that, for the foreseeable future, new genetic knowledge should be directed toward its use to prevent or cure disease. On the other hand, is there anything wrong with pursuing perfection as a goal of reproduction? Will it be part of medicine's professional responsibility to allow parents to select among traits for their future offspring? Will such choices only be available to those who are able to pay? Without serious considerations of these difficult questions today, society of the future risks being divided into those who are genetically sound and those who are genetically afflicted. There are serious inequalities in the United States in access to medical services which are correlated to both class and race. A world in which genetic testing is widely available might be incompatible with a system of private health insurance. Societies committed to equality and fairness will have to consider measures to ensure that the means to implementing eugenic choices are available to *all* who desire them. Envisaging the future world of genomics reinforces arguments for ensuring all members of society will have access to affordable medical care, so that the new technologies do not compromise the rights and aspirations of vulnerable people.

## Endnotes

1. R. Lipkin, *The Quest to Break the Human Genetic Code*, Insight 46-48 (Dec./Jan. 1991).
2. Ewan Birney, A. Bateman, M. Clam, & T. Hubbard, *Mining the Draft Human Genome*, 409 Nature 827-28 (2001).

3. The Quick Guide to Sequenced Genomes includes descriptions of these 180 organisms and has links to sequencing centers and scientific abstracts. See [http://www.genomenetwork.org/resources/sequenced\\_genomes/genome\\_guide\\_p1.shtml](http://www.genomenetwork.org/resources/sequenced_genomes/genome_guide_p1.shtml).
4. National Human Genome Research Institute, NIH News Advisory, *The Mouse Genome and the Measure of Man*, December 4, 2002, available at <http://genome.gov/pagecfm?pageID=10005831>.
5. William Shakespeare, *Hamlet*, Act II, Scene ii.
6. Jon Turney, *Frankenstein's Footsteps: Science, Genetics and Popular Culture* (New Haven: Yale University Press, 1998).
7. See Martin Brookes, *Get a Grip on Genetics*, 36 (1998).
8. *Id.*
9. Daniel Hartl & Elizabeth Jones, *Genetics, Principles and Analysis*, 4th ed. (1998).
10. Tom Wilkie, *Perilous Knowledge: The Human Genome Project and Its Implications*, 198 (1993).
11. Lori Andrews et al., *Genetics: Ethics, Law and Policy*, 6 (2002).
12. A joint Japanese-American investigation of the long-term radiation-bombing effects that was established at the direction of President Truman in 1947 to proceed with a long-term investigation of the effects of atomic radiation. See John Beatty, *Genetics in the Atomic Age: The Atomic Bomb Casualty Commission, 1947–1956*, in: K.R. Benson, J. Maienschein & R. Rainger (eds.), *The Expansion of American Biology*, 284–324 (1991).
13. See Robert Cook-Deegan, *The Gene Wars: Science, Politics, and the Human Genome*, 92 (1994).
14. ELSI's research areas are: (1) Privacy and fairness in the use and interpretation of genetic information, including prevention of misinterpretation or misuse. (2) Clinical integration of new genetic technologies, including advising on clinical policies related to genetic testing and counseling. (3) Informed consent and other research-ethics review issues related to the design, conduct, participation in, and reporting of genetics research. (4) Education on genetics and related ELSI issues for health professionals, policy-makers, and the general public. More information on ELSI is available at [www.nhgri.nih.gov/ELSI](http://www.nhgri.nih.gov/ELSI).
15. DOE representatives stressed two main reasons why the agency should play a pivotal role in the project: (1) continuity with previous large-scale and largely successful efforts in human genetics funded by the agency and its predecessors, and (2) the agency's success in developing novel technologies, together with its commitment to facilitating commercialization of new technologies through closer relations between its labs and U.S. firms. See David Galas, Testimony to Hearing Before Subcommittee on Energy Research and Development (1990), U.S. Senate, 11 July 1990, 18–19, 28–29.
16. The Genetic Privacy Act and Commentary are available at <http://www.ornl.gov/hgmis/resource/privacy/privacy1.html>.
17. Office of the Press Secretary, The White House, *Remarks by the President et al. on the Completion of the First Survey of the Entire Human Genome Project*, June 26, 2000, available at [http://clinton3.nara.gov/WH/EOP/OSTP/html/00626\\_2.html](http://clinton3.nara.gov/WH/EOP/OSTP/html/00626_2.html).
18. The International Human Genome Mapping Consortium, *A Physical Map of the Human Genome*, 409 *Nature* 934–41 (February 15, 2001); The Celera Genomics Sequencing Team, *The Sequence of the Human Genome*, *Science*, 1304–51 (February 16, 2001).
19. Francis Galton coined the term “eugenics” in 1885, defining it as the “science of improving stock—not only by judicious mating, but whatever tends to give the more suitable race or strains of blood a better chance of prevailing over the less suitable than they otherwise would have had.” See Allen Buchanan et al., *From Chance to Choice: Genetics and Justice*, 152 (2000). Galton founded the Laboratory for National Eugenics (now the Galton Laboratory) at University College, London, the first human genetics department in the world. His career also included the scientific study of fingerprints, statistical tests on the efficacy of prayer, and publication of a human-beauty map of the British Isles. Brookes, *supra* note 7, at 59.
20. Phillip Sloan, *Controlling Our Destinies: Historical, Philosophical, Ethical, and Theological Perspectives on the Human Genome Project*, 185 (2000).
21. Daniel Kelves, *Out of Eugenics: The Historical Politics of the Human Genome*, in: Daniel Kelves and Leroy Hood, *Code of Codes*, 3–36 (1992). The National Socialist Program in Nazi Germany, called *Lebensborn*, gave money, medals, housing, and other rewards to persuade “ideal” mothers and fathers to have large numbers of children in order to create a super-race of Aryan children and thus increase the representation of certain genes in the gene pool of future generations (i.e., positive population genetics). See also Daniel Kelves, *In the Name of Eugenics* (1995), first published 1985.
22. Buchanan, *supra* note 19, at 32.
23. Barbara Rothman, *The Book of Life: A Personal and Ethical Guide to Race, Normality, and the Implications of the Human Genome Project*, 58 (2001).
24. Concerned that uncontrolled immigration of “racially inferior types” would threaten the genetic health of America, many states imposed quotas on immigrants, favoring those of “better stock,” Northern and Western Europeans, while restricting “inferior” people, including Eastern Europeans, many of whom were trying to escape the more extreme racist programs of the Nazis.
25. 274 U.S. 200 (1927); see also Paul Lombardo, *Three Generations, No Imbeciles: New Light on Buck v. Bell*, 60 *N.Y.U.L. Rev.* 30, 50–62 (1985).
26. In May 2002, Virginia Governor Mark Warner apologized to the victims of sterilization for Virginia's role in the eugenics movement.
27. Matt Ridley, *Genome: The Autobiography of a Species in 23 Chapters*, 290 (1999).
28. Sloan, *supra* note 20, at 1.
29. T. Duster, *Backdoor to Eugenics* (New York and London: Routledge, 1990).
30. Jacques Loeb, *Die Umschau* 7 (1903), pp. 21, 25, quoted by Pauly.
31. According to Descartes, the goal of human knowledge and technology is that humans might become the “masters and possessors of nature.”
32. Behavior is nearly always influenced by complex sets of genes and the environment, so that the link between the immediate output of these genes and human behavior is extremely tenuous. Similarly, genetic heritage itself cannot determine in advance the content of thinking and reasoning—even if it is the prerequisite of developing these capacities. None of the large and well-funded linkage studies to date has conclusively identified a single specific gene that contributes to individual differences in behavior and what it does in the brain. See Dean Hamer, *Rethinking Behavior Genetics*, 298 *Science* 71–72 (October 4, 2002).
33. Sloan, *supra* note 20, at 92.
34. Thomas Hobbes, *Leviathan* (1651), introduction, ed. E. Curley.
35. Richard Lewontin, *It Ain't Necessarily So: The Dream of the Human Genome and Other Illusions*, 104 (2000).
36. John Harris, *Wonderwoman and Superman*, 188 (1992). “Inevitably as individuals [genetically] protected against environmental pollutants multiply, the perception of the urgency of need to eradicate such pollutants might well recede. The existence of the new breed might thus carry dangers to the environment as a whole as well as to the rest of mankind.”

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37. Preamble to the Constitution of the World Health Organization as adopted by the International Health Conference, New York, June 19–22, 1946; signed on July 22, 1946, by the representatives of 61 states (*Official Records of the World Health Organization*, no. 2, p. 100) and entered into force on April 7, 1948.
38. Stephanie Anderson, *From Chance to Choice: Genetics and Justice*, 22 J.L.M. 151 (2000) (book review).
39. *Id.*
40. *Id.*
41. Buchanan, *supra* note 19, at 161.
42. Anderson, *supra* note 38.
43. Buchanan, *supra* note 19, at 164.
44. *Id.*
45. Sloan, *supra* note 20, at 224.
46. Buchanan, *supra* note 19, at 60, 163.
47. Lewontin, *supra* note 35, at 189.
48. Sloan, *supra* note 20, at 221.
49. *Id.*
50. The Institute of Medicine, *Unequal Treatment: Confronting Racial and Ethnic Disparities in Healthcare* (2002). Racial and ethnic disparities in health care exist even when insurance status, income, age, and severity of conditions are comparable. These differences in health care occur in the context of broader historic and contemporary social and economic inequality, and persistent racial and ethnic discrimination in many sectors of American life.
51. Maxwell Mehlman et al., *Access to the Genome: The Challenge to Equality*, 87 (1998).
52. Norman Daniels, *Just Health Care*, 3 (1985).
53. Abortion is one method for preventing the birth of a child with a genetic illness or defect. Anticipating the expense of what might come to be known as more conventional gene therapies, abortion is likely to be the “primary preventive” measure that will be available to the poor and uninsured. *See* Mehlman, *supra* note 51.
54. *Id.* at 85.
55. *Id.* at 48.
56. *Id.* at 98.
57. Daniels, *supra* note 52.
58. *Id.* at 12.
59. *Id.* at 11.
60. *See, e.g.*, John Rawls, *A Theory of Justice* (1982), a theory of “justice as fairness” on which Norman Daniels’s *Just Health Care* is based. *See also* Robert Nozick, *Anarchy, State and Utopia*, 233–35 (1974), a libertarian view of entitlement to social goods.
61. During his administration, President Clinton proposed the Health Security Act legislation that would have guaranteed health insurance for virtually every American. Under pressure from health insurers and others, his effort failed and there are no plans to revive it. Clinton’s plan would not have provided blanket coverage for all health services—insureds would have been guaranteed only a package of basic health benefits.
62. *See generally* Daniels, *supra* note 52. If physical health conditions and mental abilities achievable with genetic technologies come to be regarded as necessary to enable individuals to carry out a life plan with a normal range of opportunity, then it would be unjust to deny individuals that chance.
63. Anderson, *supra* note 38, at 151.
64. Madison Powers, *Privacy and the Control of Genetic Information*, in: Mark S. Frankel & Albert Teich (eds.), *The Genetic Frontier: Ethics, Law and Policy*, 77 (1994).
65. *See generally* Daniel Callahan, *Setting Limits: Medical Goals in an Aging Society* (1995); Willard Gaylin et al., *The Perversion of Autonomy: The Proper Uses of Coercion and Constraint in a Liberal Society* (1996).
66. Wilkie, *supra* note 10, at 11.
67. *See* Graeme Laurie, *Challenging Medical-Legal Norms: The Role of Autonomy, Confidentiality, and Privacy in Protecting Individual and Familial Group Rights in Genetic Information*, 22 J. L. Med. 1–54 (2000).
68. *Id.*
69. *See*, for example, H.R. 3636, Genetic Privacy and Nondiscrimination Act of 2003, introduced to the House of Representatives on November 21, 2003. This measure is still being considered in the 109th Congress (2005–2006). The Genetic Nondiscrimination Federal Legislation Archive is available at <http://www.genome.gov/11510239>.
70. *Supra* note 15. The Genetic Privacy Act is a draft bill written in 1995 by George Annas of Boston University School of Public Health, to assist legislators in proposing new legislation in their states.
71. In March 1995 the EEOC provided some guidance by interpreting the ADA as it may relate to genetic information: “Entities that discriminate on the basis of genetic predisposition are regarding the individuals as having impairment, and such individuals are covered by the ADA.”