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## 10. GENETIC TECHNOLOGIES AND MEDICINE

### Privacy, Identity, and Informed Consent

MARSHA HANEN\*

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#### I. INTRODUCTION

In the roughly ten years since the announcement of the sequencing of the human genome, we have seen an increasing number of articles in the popular press reporting various medically related genetic breakthroughs. In recent months, we have heard claims that scientists have found a gene contributing to Alzheimer's disease, a gene for spina bifida, and a gene related to long-term memory recall, among others. There are genetic tests for certain cancers, which are, in some cases, highly controversial.<sup>1</sup> Companies aim to match the genetic makeup of individuals with pharmaceutical nutrient formulations—nutraceuticals—to enhance health and ward off disease, leading to assertions that the science of nutrigenomics is the way of the future. And we have the announcement of the sequencing of Craig Venter's personal genome,<sup>2</sup> together with urgings that others do the same, so as to create a massive augmentation of the research database. In light of all this, it is understandable if people believe that the ability to tailor medical treatment to each individual is with us already. Indeed, the

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1. See Erik Stokstad, "DNA Testing: Genetic Screen Misses Mutations in Woman at High Risk of Breast Cancer," *Science* 311 (2006): 1847.

2. See Samuel Levy, Granger Sutton, Pauline C. Ng, Lars Feuk, Aaron L. Halpern, et al., "The Diploid Genome Sequence of an Individual Human," *PLoS Biol* 5, no. 10 (2007): e254.

excitement about these developments, in the media, in the private sector, and even among researchers, universities, and funders, has been so pronounced, and exaggerated, as to be labeled “genohype.”<sup>3</sup> And much of this “genohype” is problematic, for a variety of reasons.

The work done to date on the Human Genome Project has depended upon the development of a variety of new technologies which, although they have considerable potential for leading to beneficial results, also raise legitimate concerns, many of which were outlined in the 1997 *Universal Declaration on the Human Genome and Human Rights*.<sup>4</sup> In this paper, I examine the effects of some of these emerging technologies on a range of ethical issues concerning privacy, autonomy (sometimes described as “the primary ethical value in medicine in most Western countries”<sup>5</sup>), and related aspects of the control we attempt to exercise over our lives. Specifically, I focus on these issues in relation to “personalized medicine,” commercialization, uses and misuses of information, discrimination, and informed consent. Of course, some of the concerns presented here arise in a familiar way in nongenetic medical contexts, but advances in genetics and genomics bring with them a new set of issues that are likely to multiply the concerns significantly.

## II. PERSONALIZED MEDICINE AND COMMERCIALIZATION

“Personalized medicine” is the practice of sequencing a patient’s genome and combining this information with new knowledge of the genetic basis of many diseases, as well as the genetic component of treatment. For example, in the future, sequencing a patient’s genome might reveal a predisposition for a heart condition. This information, linked with other knowledge gleaned from the patient’s genome, would allow a physician to choose the optimal prevention strategies and most effective medications for that individual. In this way, medical diagnostics and treatment might be tailored to each patient. The Personalized Medicine Coalition, an independent, nonprofit group that “works to advance the

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3. See, for example, Timothy Caulfield and Tania Bubela, “Media Representations of Genetic Discoveries: Hype in the Headlines?” *Health Law Review* 12 (2004): 53–61; Tania Bubela and Timothy Caulfield, “Do the Print Media ‘Hype’ Genetic Research?: A Comparison of Newspaper Stories and Peer-Reviewed Research Papers,” *Canadian Medical Association Journal* 170 (2004): 1399–1407.

4. Universal Declaration on the Human Genome and Human Rights, UNGA Res 53/152 (9 December 1998); also see Bartha Maria Knoppers, *Human Dignity and Genetic Heritage: A Study Paper Prepared for the Law Reform Commission of Canada* (Ottawa: The Commission, 1991); Deryck Beylveled and Roger Brownsword, *Human Dignity in Bioethics and Biolaw* (New York: Oxford University Press, 2002).

5. Dorothy C. Wertz, “Patients’ and Professionals’ Views on Autonomy, Disability and Discrimination,” in *The Commercialization of Genetic Research: Ethical, Legal and Policy Issues*, eds. Caulfield and Williams-Jones (New York: Kluwer Publishers, 1999), 171.

understanding and adoption of personalized medicine for the ultimate benefit of patients” claims that personalized medicine will transform health care by predicting likely outcomes of drug therapy and engaging in targeted drug development to improve health outcomes and increase cost-effectiveness.<sup>6</sup>

Many of these hoped-for outcomes involve commercialization of testing and genetic products. The practice of “genohype” often leads to unrealistic claims about the imminence of medical breakthroughs arising from genetic and genomic information; and the consequent trading on such information has been closely associated with a wide range of commercial activities, aimed at commodifying much of the rapidly expanding knowledge about connections between genetics and health.

The apparently great promise of personalized medicine, both for enhanced medical care and for substantial profit, has attracted a large number of companies into the field. The Personalized Medicine Coalition has a Board of Directors principally made up of large biotech/pharmaceutical companies, IT/informatics companies, diagnostic companies, health insurance companies, and venture capitalists.<sup>7</sup> And, although commercialization may lead to useful tests and treatments, it is reasonable to ask whether these companies are likely to give priority to the ethics of promoting genetic technologies or to considerations of privacy and autonomy.

These are early days for many genetic and genomic technologies, and little is known that would support the transformation of our considerable genomic knowledge into genetic therapies. Equally as important, the claims surrounding the possibilities of direct-to-consumer genetic testing for a variety of conditions bring with them little evidence that such testing is likely to provide useful results in the near term. Nevertheless, more than

1000 types of genetic tests are currently on the market for single-gene diseases, like cystic fibrosis and hemophilia. And the latest crop of DNA testing services, often sold directly to customers, offer to scan a person’s entire genome to determine the likelihood of developing more common conditions like obesity or Alzheimer’s disease.<sup>8</sup>

Critics, including many leading health policy advisors and geneticists, insist that the tests are both unproven and probably unnecessary in determining

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6. “Science and Public Policy: Personalized Medicine 101,” *Personalized Medicine Coalition*, 2007, <http://www.personalizedmedicinecoalition.org/sciencepolicy/personalmed-101-overview.php> (accessed April 20, 2007); see also Francis S. Collins, webcast, “Personalized medicine: How the human genome era will usher in a health care revolution,” *National Human Genome Research Institute*, February 10, 2005, [http://www.genome.gov/Pages/News/webcasts/Personalized\\_Medicine\\_files/Default.htm#nopreload=1](http://www.genome.gov/Pages/News/webcasts/Personalized_Medicine_files/Default.htm#nopreload=1) (accessed April 20, 2007).

7. “About PMC: PMC Members,” *Personalized Medicine Coalition*, 2007, [http://www.personalizedmedicinecoalition.org/about/pmc\\_members.php](http://www.personalizedmedicinecoalition.org/about/pmc_members.php) (accessed May 27, 2008).

8. Laura Bonetta, “Getting up Close and Personal with your Genome,” *Cell* 133 (2008): 753.

people's propensity to certain disease conditions.<sup>9</sup> And in any case, it is important to remember that genes constitute only one among many determinants of health and disease, so that the testing, even if evidence-based, still tells us little about an individual's future health.

In addition, there are numerous companies that trade on our wish to remain healthy by offering advice and products that are said to be tailored for each individual. For example, Sciona, one among many nutrigenomics companies, "provides personalized health and nutrition recommendations based on an individual's diet, lifestyle and unique genetic profile."<sup>10</sup>

Although commercialization of genomic information is inevitable and frequently useful, we need to examine and understand the various forces at work in developing both the science and its applications, including the rhetoric employed,<sup>11</sup> as a way of distinguishing possibly beneficial treatments from snake oil, and also of dealing with issues that could compromise people's autonomy and dignity.<sup>12</sup>

### III. PERSONALIZED MEDICINE, PRIVACY, AND DISCRIMINATION

Whether involving commercialization or not, several aspects of personalized medicine are already with us. Pharmacogenomics<sup>13</sup>—the study of the inherited basis of differences in response to drugs—has shown that interindividual differences in the rate that individuals metabolize medications are often more than tenfold. This means that a "slow metabolizer" or "low responsive" individual might require one-tenth of the dose of a medication recommended for a "rapid metabolizer" or "high responsive" person. The slow metabolizer is therefore more likely to experience drug toxicity from the standard prescribed dosage than

9. For example, see United States Government Accountability Office, *Testimony Before the Special Committee on Aging, Nutrigenetic testing: Tests purchased from four web sites mislead consumers: Statement of Gregory Kutz, Managing Director Forensic Audits and Special Investigations*, U.S. Senate, July 27, 2006.

10. "About Sciona," *Sciona: Optimal Health Through Genetics*, 2006, <http://www.sciona.com/> (accessed September 20, 2007).

11. Donna Haraway discusses the Human Genome Project and the claim that it is biology's equivalent to putting a man on the moon. She states "All these technoscientific travel narratives are about freedom; the free world; democracy; and, inevitably, the free market." Donna Haraway, *Modest\_Witness@Second\_Millennium.FemaleMan@\_Meets\_OncoMouse™: Feminism and Technoscience* (New York: Routledge, 1997), 167.

12. Nola Ries and Timothy Caulfield, "First Pharmacogenomics, Next Nutrigenomics: Hypohe or Genohealthy?" *Jurimetrics* 46 (2006): 281-308.

13. The term "pharmacogenomics" is used more or less interchangeably with "pharmacogenetics."

a rapid metabolizer,<sup>14</sup> although what actually happens in individual cases is a matter of probabilities rather than certainties.<sup>15</sup>

Proponents of pharmacogenetics, including leaders of the pharmaceutical industry, “. . . speak of a future in which . . . comprehensive genetic testing of individuals would become routine; large arrays of genetic data would be held for each individual; genetically targeted pharmaceuticals would reduce inappropriate prescription and (supposedly) lower drug costs.”<sup>16</sup>

However pharmacogenetics, and other genetic technologies that begin with the sequencing of a patient’s genome,<sup>17</sup> run some significant risks. If a patient’s genotype and phenotype become widely available to physicians, there is a danger that this information will be more generally disseminated, with consequent significant effects on privacy and the possibility that a variety of unauthorized or even discriminatory uses will be made of the information.

This connection between the potential for privacy violations and possible stigmatization and discrimination is a central theme of this section of the paper. A person’s autonomy and ability to direct her life and to present herself in particular ways (always within constraints over which she has varying degrees of control) is closely connected with her sense of her identity, including genetic identity, her health, and her susceptibilities to diseases. Identity, autonomy, and privacy are tied to concerns about genetic discrimination because genomic information is more sensitive than “ordinary” medical information. Genomic information has the potential to provide much larger quantities and more varied types of personal data. And, given that genetic links to certain stigmatizing diseases such as mental illnesses and addictions are being sought by researchers,<sup>18</sup>

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14. Daniel W. Nebert and Eula Bingham, “Pharmacogenomics: Out of the Lab and into the Community,” *TRENDS in Biotechnology* 19, no. 12 (2001): 519–523; For more on pharmacogenetics, also see Tom Ling and Ann Raven. “Pharmacogenetics and Uncertainty: Implications for Policy Makers,” *Studies in History and Philosophy of Science* 37, no. 3 (2006): 533–549; Abdallah Daar and Peter A. Singer, “Opinion: Pharmacogenetics and Geographical Ancestry: Implications for Drug Development and Global Health,” *Nature Reviews Genetics* 6, no. 3 (2005): 241–246; Laviero Mancinelli, Maureen Cronin, and Wolfgang Sadée, “Pharmacogenomics: The Promise of Personalized Medicine,” *AAPS PharmSci* 2, no. 1 (2000): E4.

15. Bonetta, *ibid.*, 754.

16. Onora O’Neill, “Informed Consent and Genetic Information,” *Studies in History and Philosophy of Biology and Biomedical Sciences* 32, no. 4 (2001): 689–704, 700.

17. For example, preimplantation genetic diagnosis, certain cancer and disease screenings, and some prenatal testing can include genetic testing and may, in the near future, begin with the sequencing of a patient’s genome or the genome of her fetus or embryo.

18. See for example X. Luo, H. R. Kranzier, L. Zuo, B. Z. Yang, J. Lappalainen, and J. Gelerntner, “ADH4 Gene Variation is Associated with Alcohol and Drug Dependence: Results from Family Controlled and Population-Structured Association Studies,” *Pharmacogenet Genomics* 15, no. 11 (2005): 755–768.

individuals who have their genomes sequenced now may, in the future, have to confront the possibility of discrimination if they test positive for certain gene variations.

Furthermore, discoveries of the genetic basis for certain diseases, such as multiple sclerosis or Alzheimer's, will likely translate only into an increase in the *probability* of getting these illnesses for those who are found to carry the relevant genes; but misunderstandings of what an "increased probability" means could lead some to believe that carrying certain genes means that the individual *will* get the disease. In addition, genomic information from one person can imply a great deal about the genetic makeup of her blood relatives, raising privacy concerns for those who are not even being screened for the various markers of disease. As genetic discoveries advance, stronger genetic links may be found for various diseases and more individuals may face stigmatizing results, increasing the motivation to keep genetic information private. Since widespread information sharing leads to increased privacy threats, privacy protection is critical.

One privacy concern is that, once a patient's genome sequence or the results of genetic tests become part of an individual's electronic medical file, persons other than one's medical practitioner, and those who "need to know," may have access to the information. And genetic testing is already widely entrenched in medical practice, from prenatal testing to testing of "at risk" populations. If the information thus obtained becomes available to almost anyone in, say, pharmacies, or social agencies, we may experience a loss of control with respect to information about us that we consider private.

Furthermore, the ubiquitous use of information technologies, such as data mining in relation to medical information could mean that employers, insurance companies, or others have access to the results of genetic testing.<sup>19</sup> Although complicated analysis of medical information is not new, what *is* new are the masses of data produced by genomics and DNA studies, the extent to which these studies allow us to learn things not previously available to us, and the ease with which such information can be provided technologically and connected with other information.

These concerns lead to questions about data security.<sup>20</sup> Examples abound of human errors that have led to security breaches, exposing millions of people to

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19. Data mining is a method that uses mathematical algorithms to extract implicit, previously unknown connections and patterns from large databases. The fact that the connections and patterns are largely unknown before the analysis begins can have profound ethical implications. For example, see U. Fayyad, G. Piatetsky-Shapiro, and P. Symth. "Discovery in Databases: An Overview," in *Knowledge Discovery in Databases*, eds. G. Piatetsky-Shapiro and W. J. Frawley (Menlo Park, CA: AAAI Press/MIT Press, 1996).

20. N. M. Ries and G. Moysa, "Legal Protections of Electronic Health Records: Issues of Consent and Security," *Health Law Review* 14, no. 1 (2005): 18–25.

unauthorized disclosure of their personal and medical information.<sup>21</sup> And this issue is pressing. An announcement on April 17, 2007, indicated that British Columbia “has contracted with Sun Microsystems (B.C.) Inc. and other partners to establish a province-wide electronic system of health data aimed at improving patient care and reducing medical errors.”<sup>22</sup> The project is said to be the largest in Canada, and one of the largest in North America. Advantages such as transcending the silos in which medical information currently resides, having patient-centric data, reducing the number of unnecessary tests being performed, bringing faster, more accurate diagnoses are cited. But what happens when incorrect information finds its way into the system, or when the information goes to the “wrong” place? Will there be legal or other consequences for careless handling of information or other failures so as to provide strong incentives not to allow this to happen? We must consider the sensitive nature of genetic information and recognize that personalized profiles make individuals more vulnerable to possible negative consequences. These concerns will likely increase if entire genetic profiles become a part of medical records.

There are ways to address these privacy concerns before such systems are put in place. Carnegie Mellon computer scientist Latanya Sweeney claims that there are accessible solutions to the privacy problems we face, but the solutions must be built into the systems *before* implementation. “If we build the right [privacy protecting] designs in up front, then society can decide how to turn those controls on and off. But if the technology is built without controls, it forces us to either accept the benefits of the technology without controls, or cripple it by adding them later.”<sup>23</sup>

Unfortunately, many current measures aimed at protecting genetic privacy may be falling short. Advanced coding of information may still leave it vulnerable

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21. In one instance, in March 2005, computer equipment containing the personal information of over 900,000 individuals was stolen from the American International Group (AIG), a leading international insurance organization. In January 2007, the theft of a laptop computer from a researcher’s car exposed 2900 current and former patients of Toronto’s Hospital for Sick Children to unauthorized release of their personal health information. News of the theft provoked the Ontario Privacy Commissioner to consider requiring that all such information be encrypted so as to make it more difficult to use, should it fall into the wrong hands. See Karen Howlett, “Information on 2,900 Patients Stolen with Laptop,” *The Globe and Mail*, A7, March 8, 2007.

22. Rod Mickleburgh, “Record-keeping to Leap out of Stone Age,” *The Globe and Mail*, S1, April 17, 2007.

23. Chip Walter, 2007, June 27. “Privacy isn’t dead, or at least it shouldn’t be: A Q&A with Latanya Sweeney.” *Scientific American.com*. Retrieved Sept. 6, 2007 from [http://www.sciam.com/print\\_version.cfm?articleID=6A2EF194-E7F2-99DF-3323DA6BA4346B0B](http://www.sciam.com/print_version.cfm?articleID=6A2EF194-E7F2-99DF-3323DA6BA4346B0B). See also: B. Malin and L. Sweeney, “How (Not) to Protect Genomic Data Privacy in a Distributed Network: Using Trail Re-identification to Evaluate and Design Anonymity Protection Systems,” *Journal of Biomedical Informatics* 37, no. 3 (2004): 179–192.

to hackers, and can, in any case, often be quite easily reconnected to its subjects.<sup>24</sup> A focus on coding practices aimed at protecting privacy may also ignore other important issues such as autonomy and informed consent. For example, deCODE genetics and the Icelandic government were widely criticized for coupling coding practices with policies of presumed consent when they devised biobanks for the study of population genomics.<sup>25</sup>

Rather than focusing narrowly on coding practices to protect privacy, we also need to consider the problem of genetic information falling into the “wrong” hands or being used in discriminatory ways. Legislation needs to be put in place to deal with such cases. Antidiscrimination laws are required to ensure that genetic information cannot be used by employers for hiring and firing purposes or by health insurers to prevent coverage; and additional legislation may be required to protect privacy more generally.<sup>26</sup>

The U.S. Genetic Information Nondiscrimination Act is intended to protect patients against the kind of apparent discrimination suffered by Terri Seargent in 1999.<sup>27</sup> Seargent discovered that she had alpha-1 deficiency, a respiratory disease that killed her brother. The discovery and early treatment probably saved Seargent’s life, but she was fired and lost her health coverage when her employer learned of her costly medical condition. Likewise, the Council for Responsible

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24. Malin and Sweeney, *ibid.*

25. Biobanks are depositories of stored genetic information and/or biological material for the purposes of current and future genetic research. deCODE’s database of personal medical files is commonly referred to as the Health Sector Database (HSD) (Árnason, “Coding and Consent,” 28 (n. 25)). deCODE Genetics negotiated with the Icelandic government to gain access to the medical records of Icelandic citizens, so that this information could be coded and used for research purposes. deCODE and the government argued that polls indicated that the majority of the population supported this important research and thus consent could be presumed. The personal medical information was obtained without explicit consent from patients and often without consent from medical professionals. An opt-out option was added only after citizen groups, doctors, and international organizations loudly denounced the policy. See Vilhjálmur Árnason, “Coding and Consent: Moral Challenges of the Database Project in Iceland,” *Bioethics* 18, no. 1 (2004): 27–49.

26. Some such legislation is already in place. In the United States, Congress passed the Genetic Information Nondiscrimination Act (GINA) on May 1, 2008. According to the National Human Genome Research Institute, “The act will protect individuals against discrimination based on their genetic information when it comes to health insurance and employment. These protections are intended to encourage Americans to take advantage of genetic testing as part of their medical care.” National Human Genome Research Institute. Genetic antidiscrimination bill clears congress. *Policy & ethics: Critical issues and legislation surrounding genetic research*. Retrieved May 15, 2008 from <http://www.genome.gov/PolicyEthics/>.

27. Diane Martindale, “Genetics Discrimination: Pink Slip in your Genes,” *Scientific American*, January 284, no. 1 (2001): 19–20.

Genetics<sup>28</sup> reported that, when genetic testing revealed that a young boy had fragile X syndrome, “an inherited form of mental retardation,” the family’s insurance company dropped the boy’s health insurance “claiming that his disability represents a preexisting condition.”<sup>29</sup> In another reported case, a social worker lost her job within a week of mentioning that her mother had died of Huntington’s disease.<sup>30</sup> And concerns about genetic discrimination are not unique to the United States.<sup>31</sup>

Genetic discrimination can have far-reaching social consequences that go beyond implications for individuals and their families. The Council for Responsible Genetics discusses a case where a lead battery manufacturing operation attempted to bar women from working for the company because lead interferes with reproductive capabilities.<sup>32</sup> “One of the main occupational health aspects of this is that employers would rather discriminate against prospective employees than clean up the work place.”<sup>33</sup> Indeed, the Council argues that it is actually not acceptable to expose anyone—men, or sterilized women, for that matter—to these excess lead levels.

The Council also raises the concern that genetic susceptibilities might be seen as “preexisting conditions” that could be used to “justify” workplace discrimination. But if this is our future, they claim:

(1) We are treating those people as damaged goods. We are devaluing their personality. (2) We are discriminating in ways that are not justifiable. The employers should clean up their act.<sup>34</sup>

Furthermore, genetic discrimination in this area will likely serve to further disadvantage those who are already marginalized. And the issue of marginalization

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28. Council for Responsible Genetics, “Genetic Discrimination: A Position Paper Presented by the Council for Responsible Genetics,” (original version written 1997; January 2001), <http://www.gene-watch.org/programs/privacy/genetic-disc-position.html> (accessed April 20, 2007).

29. *Ibid.*

30. *Ibid.* In this example, the social worker has a 50% chance of carrying the gene that causes Huntington’s disease. Unlike most diseases with a genetic component, Huntington’s is a genetically inherited disease. It only takes one copy of the HD gene to cause Huntington’s disease (it is an autosomal dominant trait). Persons who are gene carriers *will* develop the disease (assuming that they live long enough).

31. See, for example, Trudo Lemmens, Mireille Lacroix, and Roxanne Mykitiuk, *Reading the Future? Legal and Ethical Challenges of Predictive Genetic Testing* (Montreal: Les Éditions Thémis, 2007).

32. Diane Horn, interview with Phil Bereano, “Genetic Discrimination: A Primer,” *Council for Responsible Genetics*, KCMU 90.3 FM-Radio, Seattle, Washington, <http://www.gene-watch.org/programs/privacy/BerInterview.html> (accessed April 24, 2007).

33. *Ibid.*

34. *Ibid.*

is particularly important in the context of discrimination as it is the poor, the homeless, immigrants, and aboriginal people who are most likely to be subject to discrimination.

#### IV. GENETIC TECHNOLOGIES, IDENTITY, AND AUTONOMY

The focus of the paper on autonomy and related issues that have an ethical dimension is crucial because of the importance of viewing persons in medical contexts as “possessing dignity and inherent worth”<sup>35</sup> and being able to make rational choices for their lives. But because of the problem of marginalization, our view of autonomy must go beyond the purely individualistic. We need a more contextualized, socially situated, “relational” notion, which “examines patient autonomy in the social and political dimensions within which it resides and provides us with the theoretical resources that we need for restructuring health care practices in ways that will genuinely expand the autonomy of all patients.”<sup>36</sup> We need to take account of the factors that influence our ability to exercise our autonomy and maintain our sense of identity based on social, as well as the traditional individual understandings of health and the factors that affect it.

There are also serious ethical implications for autonomy and identity stemming from misconceptions that many diseases are “purely” genetic in origin. The focus on genetics and disease sometimes leads to a kind of reductionism—“genetic essentialism” or “genetic determinism,”<sup>37</sup> which holds that our genes determine almost everything about us, to the exclusion of other important influences.

Actually, genetic and environmental interactions mean that few diseases are “purely” genetic in character. For example, a person whose birth is extremely premature may develop chronic respiratory illnesses that do not show up on her genetic map; likewise, people who work in coal mines, or have been in serious car accidents may develop illnesses unrelated to their genetic makeup. Carriers of the “breast cancer genes” have an increased risk for developing breast cancer,

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35. Kwame Anthony Appiah, *The Ethics of Identity* (Princeton, NJ: Princeton University Press, 2005), 61.

36. Susan Sherwin, *The Politics of Women's Health: Exploring Agency and Autonomy* (Philadelphia, PA: Temple University Press, 1998), 44.

37. There are numerous examples in the literature of the problems associated with accepting genetic determinism. See for example, Dorothy Nelkin and Susan M. Lindee, *The DNA Mystique: The Gene as a Cultural Icon* (Ann Arbor: University of Michigan Press, 2004); R. C. Lewontin, Steven Rose, and Leon J. Kamin, *Not in Our Genes* (New York: Pantheon Books, 1984); Celeste M. Condit and Deirdre M. Condit, “Blueprints and Recipes: Gendered Metaphors for Genetic Medicine,” *Journal of Medical Humanities* 22, no. 1 (2001): 29–39. See also Abby Lippman, “The Politics of Health: Geneticization Versus Health Promotion,” in Sherwin, *The Politics of Women's Health* (1998): 64–82.

but this does not mean that they will inevitably develop the disease, nor does it mean that those who do not carry these genes will not develop it. So, although current genetic research may appear to suggest that we are on the verge of understanding, and even eradicating many major diseases, it is important to keep in mind that “genetic status” gives us only limited prediction of future illness;<sup>38</sup> and overemphasizing genetic makeup could negatively affect people’s view of themselves and their sense of their own autonomy.

Furthermore, and contrary to the expectations of many, information about connections between genes and particular diseases has generally not led to breakthroughs in treatment.<sup>39</sup> If important environmental factors and the complexity of disease are overlooked, conclusions can be seriously misleading and even damaging; it is not unreasonable to be concerned that the medical profession may pay less attention to environmental causes of disease as more information about the genome comes to light and a “geno-centric” view of disease becomes entrenched.<sup>40</sup>

Some have worried that genetic technologies and the push for personalized medicine could lead to a loss of autonomy or confusion about identity because we do not have a clear conception of what “disease” and “wellness” mean. As S. O. Hansson notes,

Disease is not a biologically well defined concept but one that depends largely on social values. Some conditions previously regarded as diseases are now thought of as normal states of the mind or body. Others that were previously perceived as variations of normality are now regarded as diseases. Homosexuality is an example of the former, attention deficit hyperactivity disorder of the latter.<sup>41</sup>

Confusion about medical terminology is compounded by confusion about the role of genetic technologies: Will the focus be to cure, to prevent disease, to screen for and eliminate genetic “abnormalities,” or will the focus switch to “enhancement”

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38. Maxwell J. Mehlman and Jeffery R. Botkin, *Access to the Genome: The Challenge to Equality* (Washington, DC: Georgetown University Press, 1998), 25.

39. The National Human Genome Research Institute states, “It is important to realize . . . that it often takes considerable time, effort, and funding to move discoveries from the scientific laboratory into the medical clinic. Most new drugs based on genome-based research are estimated to be at least 10 to 15 years away. According to biotechnology experts, it usually takes more than a decade for a company to conduct the kinds of clinical studies needed to receive approval from the Food and Drug Administration.” “A Brief Guide to Genomics,” *National Human Genome Research Institute*, June 27, 2007, <http://www.genome.gov/18016863> (accessed September 21, 2007).

40. Recent research suggests that there may be a stronger link between gene expression and environmental factors than previously thought. We are beginning to see a biological basis for environmental influences and social determinants of health through epigenetics—the study of the link between gene expression and environmental factors. This could eventually lead to a clearer understanding of individual genomes and individual susceptibilities.

41. S. O. Hansson, “Implant Ethics,” *Journal of Medical Ethics* 31 (2005): 519–525; 522.

and creating longer, better lives for people? Much depends upon how “abnormalities” are defined and viewed, and whether these definitions will change our view of our own identities and ability to make autonomous decisions about treatment. We need also to ensure that access to new technologies will not be a function of wealth or influence rather than need, or other more egalitarian criteria.

In a context in which people are increasingly being viewed as “responsible” for many of their illnesses, because of what are viewed as poor “lifestyle” choices—inappropriate diet, lack of exercise, smoking, and the like—the belief easily develops that individuals have the power to maintain optimal health, regardless of poverty, homelessness, mental illness, or environmental toxicities.<sup>42</sup> Similarly, if the expectation develops that technologies available for preventing or removing genetic “abnormalities” will be used in ways advocated by professional or corporate interests, who will also determine which people “deserve” care, there is a significant risk of unfairness and dysfunction in the system.

P. J. Boyle correctly notes that genetic research is neither more “neutral” than much other scientific research, nor are researchers “mere spectators to the unfolding of the secrets of the human genome.”

We plant the seeds of the answers we will arrive at in the way we frame the questions we ask. The genetic knowledge we shape in such a manner will in turn dictate the nature of our social, legal, and ethical responsibilities.<sup>43</sup>

Likewise, genetic technologies give rise to questions of ethical responsibility for physicians. What should be the role of the practitioner in situations where a patient could lose her health insurance, her job, or be exposed to other forms of discrimination as a result of genetic findings? If a woman requests genetic testing for breast cancer genes and the genes are found, she may be vulnerable to genetic discrimination, whether or not she ever develops cancer. And where testing could have a negative impact on a patient’s career, position in society, or relationships with other people it is reasonable to ask whether the physician has a duty to warn the patient of such potential impacts. On the other hand, we might ask whether we should assume that the patient has a right to know, and that the negative consequences to patient autonomy always tell against attempting to protect a patient from worrisome knowledge.

Medical professionals must also take account of their possible responsibilities to the patient’s relatives when genetic testing is involved. Consider the example

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42. Researchers working in epigenetics are showing how our “lifestyles and environment can change the way our genes are expressed, leading even identical twins to become distinct as they age.” NOVA: Science Now. (PBS) 2007, July. *Epigenetics*. Retrieved Sept. 21, 2007 from <http://www.pbs.org/wgbh/nova/sciencenow/3411/02.html>. For more on the science of epigenetics, see A. Bird, “Perceptions of Epigenetics,” *Nature* 447 (2007): 396–398.

43. P. J. Boyle et al., “Genetic Grammar: Health, Illness, and the Human Genome Project,” *Hastings Center Report*, Special Supplement 22, no. 4 (1992): S1.

of the woman whose mother died of medullary cancer.<sup>44</sup> Three years later the woman was diagnosed with the same cancer, already in an advanced stage. She felt that her mother's doctor should have disclosed to her the fact that the disease is transmitted genetically as a dominant trait and sued the doctor for not passing on information about her mother's genetic makeup that affected the woman herself. "The Florida Court ruled that, in the usual doctor-patient relationship, the physician has no legal obligation to speak with other members of the family about their risks."<sup>45</sup> But, from a moral point of view, perhaps a different conclusion would be in order, especially in light of the effect the physician's decision may have had on the daughter's autonomy and sense of self.

Also consider instances where individuals who have had a grandparent die of Huntington's disease have chosen to be tested for Huntington's even though their asymptomatic parent has chosen not to be tested. If the individual tests positive for the Huntington's allele, then the parent is a carrier and will develop the disease. In these cases the parent is likely to find out something about his or her own genetic makeup that he or she did not want to know. Such scenarios could become more common as genetic technologies advance and more diseases with genetic origins are discovered, even in the case of diseases not as deterministic as Huntington's. Does a physician have any duty to individuals who are not patients if the information also pertains to them?

These examples highlight how the identity and autonomy of the involved parties can conflict with one another: One person's autonomous choice to learn facts about herself could change the way another person views himself. In the first of these examples, the daughter was deprived of information to which she felt entitled, whereas in the second, the parent is likely to gain information about his own genetic makeup that he does not wish to have. The physician must grapple with the ethical concerns of her patient as well as considering the ethical questions that arise from acquiring critical medical information about *another* person. These examples also help to illustrate the complexity of issues surrounding genetic testing, and point to ways in which genetic technologies complicate the informed consent process.

## V. INFORMED CONSENT AND TRUST

Onora O'Neill writes,

*Informed consent* has been seen as the key ethical requirement for medical treatment and research, to be supported by requirements for *professional confidentiality* and for *personal privacy*. Securing the informed consent of patients

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44. Nebert and Bingham, 2001, p.521 (n. 14).

45. *Ibid.*, 521.

and respecting the confidentiality of information they provide have been seen as operationalising the ethical ideals of respecting individuals, their rights and their autonomy.<sup>46</sup>

Among the reasons for a systematic insistence on informed consent in medical contexts is society's belief in the importance of people's ability to make autonomous decisions about what they wish to keep private about themselves, and about the identities that they wish to project to the world. In this context, the issue of trust looms large, particularly with respect to consent to medical procedures, research, and other uses of genetic information. Trust and trustworthiness underline concerns about security of information, genetic discrimination, and even the usefulness and appropriateness of genetic testing itself, and trust is clearly tied to the practice of informed consent in that the patient needs to be able to trust that her wishes with respect to her genetic information will be honored.

Yet it is often difficult, when consent is sought, to be certain just what is being consented to. Much depends upon how procedures or research or other processes are described to the person whose consent is needed, and how much contextual information as well as overall understanding that individual possesses.

In particular, because a considerable portion of most patients' knowledge of medical matters derives from what they are told by their physicians, and people not medically trained may have difficulty understanding some of what they are told, trust becomes more necessary, but harder to sustain. In terms of informed consent, this difficulty is summarized by O'Neill:

Consent is particularly problematical in medical practice, because it is commonplace even for patients who are in the maturity of their faculties to find themselves at a time of weakness and distress surrounded by others who seem (and may be) more knowledgeable, whose influence and power are considerable, whom they very much do not want to offend. If consent is to be a governing principle in medical ethics, we seemingly need to be ideal rational patients; but when we are patients we are often furthest from being ideally rational . . .<sup>47</sup>

Moreover, the more complicated the technology being brought to bear, the more likely it is that patients will have limited knowledge with which to make decisions, so it is easy to see how dependent we become on trusting the practitioner.

We must also consider that in the medical context, it is not unusual for people who live in "situations of oppression, marginality, illiteracy, poverty, or a range

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46. Onora O'Neill, "Informed Consent and Genetic Information," *Studies in History and Philosophy of Biology and Biomedical Sciences* 32, no. 4 (2001): 689–704; 691.

47. *Ibid.*, 693.

of other"<sup>48</sup> circumstances to find it impossible, without the help of an advocate of some sort, to take charge of their own medical decisions in the face of physician expertise that is, frequently, experienced as intimidating. Therefore, although much of the discussion of physician-patient relationships assumes that patients are fully autonomous beings, this view is unrealistic in many cases. This is important, because informed consent is supposed to enable individuals to make autonomous decisions about what they wish to keep private about themselves, and for marginalized members of society, "sensitive information" may be even more "sensitive," as there may be more at stake if their privacy is threatened.<sup>49</sup>

These worries can be translated into a more general critique of the informed consent process. As O'Neill writes,

Consent is a *propositional attitude*: it is always directed to some description of a proposal, situation or action. Its object is always some specific *propositional content*. Where a proposition consented to misdescribes a proposed action, or is economical with the truth, consent may be misdirected and so will not be legitimate. This is all too common . . . The ethical implications of the *referential opacity of propositional attitudes* are massive. We generally consent in the required, informed and freely chosen way to rather little: so rather little can be legitimated by appeal to consent.<sup>50</sup>

Genetic technologies compound the opacity of the informed consent process because of the immense amount of information that comprises genetic decision making. O'Neill claims that it is not merely that science has made these decisions more complex:

We remain finite, ignorant and vulnerable agents with limited cognitive capacities, limited abilities to choose and limited time: but in medical contexts we face, and will increasingly face, vastly complex ranges of information, organised in the increasingly formalized ways demanded by increasingly intricately structured regulatory processes. Nowhere is this more evident than in those parts of medicine and of life which are most affected by the increasing complexity and availability of genetic data, and by the increasing variety of ways in which such knowledge may be collected, stored, used and disclosed.<sup>51</sup>

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48. Lorraine Code, *Ecological Thinking: The Politics of Epistemic Location* (New York: Oxford University Press, 2006), 187.

49. For example, individuals in lower socio-economic situations may be more vulnerable to the genetic discrimination that could result from employers exploiting links between various environmental factors and genetics, such as the example of the battery factory. Diane Horn, interview (n. 32).

50. O'Neill, "Informed Consent and Genetic Information," 692, (n. 46).

51. *Ibid.*, 695.

Thus, O'Neill believes that our cognitive capacities are being overwhelmed and more attempts to make decisions easier and more straightforward will make the process even more opaque. O'Neill's critique highlights another important consideration. In our technological society, with genetics so often featured in the media, it is hard to know what expectations are reasonable to set with respect to becoming informed. Often we feel that we should know more about DNA, genetics, and the human genome, not to mention data mining and other information technologies. These high expectations may make patients feel responsible and inadequate in decision making situations, and they may be reluctant to ask questions or to delay consenting to procedures, particularly in clinical environments where formal consent is often sought at the last moment, when there is little time available for discussion. Furthermore, it is not just what a patient knows (or does not know) but also how much *is known*. As indicated earlier, the way that genetic technologies will shape our future and the associated privacy implications are largely *unknown* at this time. Processes based on the notions of informed consent must make these points transparent.

O'Neill notes that in a typical biomedical setting autonomy and informed consent are given center stage, while trust is pushed to the margins. But she takes the position that trust is more important than autonomy "in any ethically adequate practice of medicine, science and biotechnology."<sup>52</sup> As Daniel Callahan argues, autonomy "is *a* value, not *the* value," and proper medical practice requires "a search for morality in the company of others, community as an ideal and interdependence as a perceived reality, and an embracing of autonomy as a necessary but not a sufficient condition for a moral life."<sup>53</sup>

The use of genetic technologies may be especially damaging to the trust necessary in medicine and research. Medical care that is specific to the individual's genetic profile may appear to some to be the ultimate in personalized medicine: it may look as though the medical practitioner has come to know you intimately and is designing care with your unique characteristics in mind. However, this is more appearance than reality. It is unlikely that the intimate and individual doctor-patient relationship that appears as the paradigm of good medical practice will be realized in this genetic age. And it is reasonable to ask whether it should be. It might be more appropriate to maintain that transparency about medical procedures and informed consent processes are the only things that can

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52. Onora O'Neill, *Autonomy and Trust in Bioethics* (Cambridge: Cambridge University Press, 2002), ix.

53. Daniel Callahan, "Autonomy: A Moral Good, Not a Moral Obsession," *The Hastings Center Report* 14, no. 5 (1984): 40–42; see also Willard Gaylin and Bruce Jennings, *The Perversion of Autonomy: Coercion and Constraints in a Liberal Society* (Washington, DC: Georgetown University Press, 2003).

solidify trust between individuals and the medical professionals with whom they come in contact.

Furthermore, it may be that we need to think in new ways about informed consent itself. O'Neill, for example, believes that, because of the extreme quantity and complexity of genetic information that would have to be grasped in each instance of granting consent, the emphasis in the future will have to be on constructing trustworthy institutions rather than on individual acts of consent; and such institutions will only earn their designation as trustworthy if "there are feasible procedures by which an individual can check on what is done."<sup>54</sup> But whether it is at all practical to create such institutions remains to be seen, so it would, at this stage, be unwise to be too sanguine about the possibility of finding techniques adequate to sustaining trust and giving voice to people's apparent need to maintain their ability to control the use of their genetic information.

## VI. CONCLUSION

Genetic technologies and genomic information are rapidly evolving, and enormous changes can be expected in the next few years. Clearly, there is tremendous promise in genetic medicine—only a small part of which has been realized to date. There are also potential pitfalls, which have been much discussed, but have not been dealt with in an integrated way.

The increased use of genetic information in medical contexts raises questions about who decides on the collection of genetic material and applications of genetic technology and what safeguards need to be in place to guard against errors of fact or interpretation and poor decisions that could be harmful to individuals or groups. In the varied and rapidly changing landscape of medical knowledge, the availability of reliable sources of information to medical decision makers would go a long way toward raising people's confidence that their genetic information will be used in their best interests. In order to deal appropriately with the issues of privacy and the ways in which genomics has the potential to make us think differently about our identities, we must recognize that we need much more information, and time to think carefully, not only about people's genomes, but also about how genomic information interacts with environmental characteristics. However tempting commercial applications may be, we need to assess both their positive and negative implications, especially the possible effects on privacy, autonomy, dignity, and even people's sense of who they are. And where a need for policies or laws

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54. O'Neill, "Informed Consent and Genetic Information," 702–703 (n. 46).

to protect privacy is identified, appropriate methods of enforcement and measures of accountability must be included.

In all of this, informed consent plays a major role, because it represents people's ability to make autonomous decisions about their lives. Such decisions may be different for different people, so it is important that the framework within which decisions are made allows for such variations, and recognizes that the groups to which people belong, whether through their choice or not, play a significant role in how their medical care will develop.