Chapter 7

Genetic Privacy

George Annas

The human genome project has brought with it many legal and ethical issues, but the most consistently contentious is genetic privacy.[1] As DNA sequences become understood as information, and as this information becomes easier to use in digitalized form, public concerns about internet and e-commerce privacy are merging with concerns about medical record privacy and genetic privacy. Privacy has returned to the center of American domestic public policy.

Privacy

Privacy is a complex concept involving several different but overlapping personal interests. It encompasses informational privacy (having control over highly personal information about ourselves), relational privacy (determining with whom we have

personal, intimate relationships), privacy in decision-making (freedom from the surveillance and influence of others when making personal decisions) and the right to exclude others from our personal things and places. In the U.S., no single law protects all of these interests, and privacy law refers to the aggregate of privacy protections found in constitutions, statutes, regulations and common law. Together these laws reflect the value that U.S. citizens place upon individual privacy, sometimes referred to as "the right to be left alone" and the right to be free of outside intrusion, not as an end in itself, but as a means of enhancing individual freedom in various aspects of our lives. This centrality of individual freedom in the health care context is evident in state laws that establish a patient's right to make informed choices about treatment, that place an obligation on physicians to maintain patient confidentiality, and that regulate the maintenance of medical records.[2]

Privacy laws in the U.S. are fragmented because of the multiple sources of law, including the federal

government and all 50 states. Legislation is also often the result of negotiated agreements among segments of a diverse, pluralistic, and oftentimes polarized society, rather than of a real consensus. This is perhaps most readily seen in the rules that govern highly sensitive and personal data in the U.S. Unlike the approach of the European Data Protection Directive, which establishes similar rights and duties relative to different kinds of personal data (health and finance), the U.S has different rights and duties for personal information depending upon the kind of information involved.[3] There are even different rules for different types of information in medical records. For example, the U.S. has laws that govern medical record information generally, as well as separate laws that govern specific types of medical information, such as HIV status, substance abuse treatment information, and mental health information. New federal regulations will apply the same privacy rules to all medical information except psychotherapy notes.[4] Such exceptionalism has been criticized. The primary argument against specific laws designed to protect

genetic information is that "genetic exceptionalism" would perpetuate the misconception that genetic information is uniquely private and sensitive.[5]

Genetic privacy

Is DNA sequence information uniquely private, or is it just like other especially sensitive information contained in an individual's medical record? If it is not unique, existing medical record confidentiality laws should be sufficient to protect genetic sequence information, and no new laws would be needed. Those who support genetic exceptionalism emphasize the unique distinguishing features of DNA sequence information. The DNA molecule itself is a source of medical information, and like a personal medical record, it can be stored and accessed without the need to return to the person from whom the DNA was collected for authorization. But DNA sequence information contains information beyond an individual's medical history and current health status. DNA also contains information about the

individual's future health risks, and in this sense is analogous to a probabilistic coded "future diary."[6] As the code is broken, DNA reveals information about an individual's probable risks of suffering from specific medical conditions in the future. A number of commentators have noted how private and personal diaries are, and why they should be treated with unique respect. William Safire, for example, has argued that we keep our diaries "to reveal our youthful selves to our aging selves."[7] The DNA molecule is the converse of that: the decoded DNA molecule reveals our aging selves to our younger selves. Of course it's probabilistic not deterministic; and it's in a code that we are only in the process of breaking. At some point, however, we as scientists are going to be able to read our DNA and tell us something about the types of diseases that we are at risk to encounter as we age. There is nothing else quite like this type of information.

Our current obsession with genetic sequence information also means that it is likely to be taken more seriously than other information in a medical

record that could also predict future risks, like high blood pressure or cholesterol levels. Information about the presence of proteins that specific genes may code for is also different from DNA sequence information because their presence may change over time, and their levels, like cholesterol readings, can only be determined over time by retesting the patient personally. DNA sequence information is stable and remains the same. In contrast, proteomics (the search for all the proteins our genes code for) is more like cholesterol levels, and will not require new privacy rules, but rather enforcement of existing medical records privacy rules. DNA sequence information may also contain information about behavioral traits, such as a propensity to violence, that are unrelated to health status, although significant skepticism is called for in this area.[8]

My use of the future diary metaphor has been criticized as potentially perpetuating a mistaken deterministic view of genes.[9] I understand this criticism, and also reject the idea that genes alone or even primarily determine our future. Nurture

matters mightily. Nevertheless, I continue to believe the future diary metaphor best conveys the private nature of genetic information itself. Our future medical status is not determined solely by genetics, any more than our diaries are the only source for accurate information about our past. The DNA information, like the diary, however, is a uniquely private part of our possible future. Moreover, an individual's DNA can also reveal information about risks and traits that are shared with genetic relatives, and thus has been used to prove paternity and other relationships. DNA has the paradoxical quality of being unique to an individual, yet shared with others.[10]

Finally, even a conclusion that DNA sequence information (derived from analyzing the DNA molecule) is no more sensitive than other medical information, tells us nothing about the need to protect the DNA molecule itself. In this regard, it is useful to view the DNA molecule as a medical record in its own right for privacy purposes. Possessing a DNA sample from an individual is analogous to having medical information

about the individual stored on a computer disk, except with DNA the information is stored in a blood or other tissue sample. Like the computer disk, the DNA sequence can be "read" by the application of technology. Thus, regardless of the rules developed to control the use of genetic information that is recorded in traditional paper and electronic medical records, separate rules are needed to regulate the collection, analysis, storage and release of DNA samples themselves. This is because once a physician, researcher or police investigator has a DNA sample, there is no practical need for further contact with the individual from whom the DNA was obtained, and additional DNA tests could be done on the stored sample (and thus on the individual) without authorization. Some of these tests are, of course, not yet developed, but all will produce new genetic information about the individual.

DNA has also been culturally endowed with a power and significance exceeding that of other medical information.[11] Much of this significance is undoubtedly misplaced. Nonetheless, it can be justified in so far

as genetic information can radically change the way people view themselves and family members, as well as the way that others view them. The history of genetic testing, particularly in relation to rare monogenic diseases such as Huntington disease, provides us with examples of this impact. Studies of individuals who have undergone testing in clinical settings demonstrate changes in self-perception caused by positive, as well as negative, test results.[12] Individuals with decreased risk of having a genetic disease have reported difficulty in setting expectations for their personal and professional lives in a more open-ended future. Adjustments appear to have been particularly difficult for those who previously had made reproductive decisions on the presumption that they were at high risk for developing a disease. Consequently, it is good public policy to provide genetic counseling before and after testing. To protect the privacy of children and adolescents, some institutions have also adopted a policy of refusing parental requests to test children for late

onset diseases when no medical intervention is available to prevent or alleviate the disease.[13]

Only one U.S. court has squarely addressed whether constitutional rights to privacy are implicated by genetic testing. In *Norman-Bloodsaw v. Lawrence Berkeley Laboratory*, employees of a research facility owned and operated by state and federal agencies alleged that nonconsensual genetic testing by their employers violated their rights to privacy. Holding that the right to privacy protects against the collection of information by illicit means as well as unauthorized disclosures to third parties, the court stated: "One can think of few subject areas more personal and more likely to implicate privacy interests than that of one's health or genetic make-up."[14]

Ownership of DNA

In Ralph Nader's brief presidential crusade for the Green Party, the line that he usually got the heaviest applause for was "our genes are not for sale." That's

another way to say that we own our bodies. And the consequence is that no one should be able to take our DNA without our permission, and no one should be able to sell it and commercialize it without our agreement.

The question of ownership of DNA is very important, and in the U.S. we haven't really confronted it yet. The Genetic Privacy Act, which my colleagues Leonard Glantz and Winnie Roche drafted for the Ethical, Legal and Social Implications program of the federal Human Genome Project, provides that individuals own their own DNA, and that no one else can use your DNA without your authorization.[15] On the other hand, existing state statutes on genetic privacy do not so provide. Instead they implicitly follow the lead of the John Moore case, in which the California Supreme Court held that even though a physician had sold a cell line derived from John Moore's spleen to a private biotech company without his permission, John Moore could claim no property interest in his cells.[16] Nonetheless, the legal position that everybody but the individual from whom DNA is extracted can own DNA is not sustainable. Either no one should be permitted to

own and sell DNA, or individuals should have property rights to their own DNA.

Acknowledging property interests in DNA need not impede research anymore than respect for individual privacy would. To the contrary, individuals are free to grant researchers property rights in their DNA, and are much more likely to do so if their privacy can be guaranteed (as it can be if identifiers are not retained). The real issue is control over the private information contained in your DNA, and ownership is the traditional way to describe and conceptualize control.

DNA research and privacy

Since the human genome was roughly sequenced, attention has shifted to research on genetic variation designed to locate genes and gene sequences with disease-producing or -prevention properties. Some researchers have already taken steps to form partnerships and create large DNA banks that will furnish the material for this research. Others want to

take advantage of the large number of stored tissue samples that already exist. In the U.S., for example, the DNA of about 20 million people is collected and stored each year in tissue collections ranging from fewer than 200 to more than 92 million samples.[17] Collections include Guthrie cards on which blood from newborns has been collected for phenylketonuria screening since the 1960s, paraffin blocks used by pathologists to store specimens, blood bank samples, forensic specimens, and the U. S. military's bank of samples for use in identifying bodily remains. Perhaps the major reason why neither DNA sequence information nor DNA samples themselves have been afforded special privacy protection is the strongly-held view of many genetic researchers and biotechnology companies that privacy protections would interfere with their work.

Several factors have contributed to the proliferation of DNA banking: the relative ease with which DNA can be collected, its coincidental presence in bodily specimens collected for other reasons, and its immutability. Regardless of the original purposes for storing specimens, however, as the ability to

extract information from DNA increases and the focus of research shifts to genetic factors that contribute to human diseases and behaviors, repositories containing the DNA of sizeable populations can be "gold mines" of genetic information. Thus it is not surprising that there is considerable interest on the part of biomedical researchers, companies that market genomic data, and the pharmaceutical industry to stake claims on these informational resources and to exploit them for their own purposes.

Commercial enterprises, as well as academic researchers, have equally strong interests in making it relatively easy to get access to DNA samples that can be linked to medical records for research purposes. Representatives of these constituencies have been vocal in arguing that requirements for informed consent and the right to withdraw data from ongoing research projects (two aspects of genetic privacy) would greatly hamper their research efforts.[18] When U.S. federal rules apply to such research—as is the case with federally-funded projects and any projects related to obtaining Food and Drug Administration

approval to market drugs or devices—a local Institutional Review Board (IRB), mandated by federal law and made up primarily of other researchers, must approve the research protocol and the informed consent process. I do not believe IRBs should waive basic federal research requirements on informed consent for DNA-based research (nor exempt researchers from them) except when the IRB determines that the research will be conducted in such a way that the subjects *cannot* be personally identified. Only when identification of individuals is impossible is there no risk to their privacy.

The most internationally discussed DNA-based project has been deCODE in Iceland, a commercial project which has been opposed by the Iceland Medical Association, among others, for ethical shortcuts, including "opt out" provisions instead of requiring informed consent of subjects.[19] The deCODE project, which has been endorsed by two acts of the Iceland parliament, involves the creation of two new databases: the first containing the medical records of all Iceland citizens, and the second DNA samples from

them (a third database, of genealogical records, already exists). deCODE intends to use these three databases in various combinations to seek out genetic variations that could be of pharmaceutical interest. The major ethical issues raised by this project are (1) the question of informed consent for inclusion of personal medical information in the database, which is currently included under the concept of presumed consent (which requires individuals to actively opt out of the research if they do not want their information in the database); (2) informed consent for the inclusion of DNA in the DNA databank in an identifiable manner (whether encrypted or not, and no matter which entity holds the encryption key); and (3) whether the right to withdraw from the research (including the right to withdraw both the DNA sample itself from the databank and all information generated about it) can be effectively exercised. Other issues include the security of the databases, and community benefit from the research project itself. Iceland provides a type of ethical laboratory that helps identify the major issues involved in population-based

genetic research, as well as helping to inform us as to why international privacy rules are desirable.

Although Icelanders themselves do not seem overly concerned with the adequacy of deCODE's plans to protect their personal privacy, other countries have been less disposed to legislating away the autonomy and privacy of their citizens. Both Estonia and the U.K., for example, have announced that their population-based DNA collections and research projects will contain strong consent and privacy-protection provisions. The privacy problems inherent in large population-based projects could be avoided altogether by stripping DNA samples of their identifiers in a way that makes it impossible to link personal medical information with DNA samples (at least by using standard identifying methods). Of course, most researchers want to retain identifiers to do follow-up work or confirm diagnoses.[20] Such identification retention, however, puts individuals at risk for breach of confidentiality and invasion of privacy, and these risks are why both informed consent and strong

privacy protection protocols are ethically necessary for genetic research.

These considerations also apply to forensic DNA databases, since even convicted felons have privacy rights, including the right not to be used as research subjects without consent.[21] DNA is collected in the forensic setting to be used for identification purposes, much like a finger print, which is why it is sometimes referred to as a "DNA fingerprint." This use is legitimate, but it does not give law enforcement officials unfettered dominion over the use of DNA. Even prisoners have a right not to have their DNA used for research purposes without specific, informed consent, and IRB review of the research protocol.[22] I also believe it is virtually impossible to obtain voluntary consent for this type of research for prisoners because they are in an inherently coercive environment, and are thus not free to refuse. What type of DNA-based research might a prison system or the FBI want to conduct? The most potentially dangerous type of disease is looking for a criminal gene, a gene that predisposes people to violence. I

don't think it exists, but whether it exists or not opens the door to labeling people and then using the genetic label to viciously discriminate against them. And of course if a researcher studies just the DNA samples found in criminal banks, the researcher is likely to find some associations. Just as XYY syndrome was used by defense attorneys to say that men with XYY are more likely to be criminals. The answer was simply that there are more XYY men in jail than there are in the free-living population. It doesn't show a cause and effect relationship, it just shows an association that may or may not be a cause and effect relationship, but in the case of XYY seems not to be. An alternative hypothesis, for example, is simply that XYY men are taller than others, and so more likely to be seen or identified during or after a crime.[23]

Risks of disclosure of personal genetic information are so high that some prominent genetic researchers, including Francis Collins and Craig Venter, have suggested concentrating not on privacy rules, but instead on anti-discrimination legislation designed to protect individuals when their genetic

information is disclosed, and insurance companies, employers, or others want to use that information against them. In June 2001, President Bush indicated that he agreed, and said he would support federal genetic antidiscrimination legislation.[24] Antidiscrimination legislation is desirable, but it does not substitute for privacy rules that can prevent the genetic information from being created in the first place without the individual's informed authorization.

A law recently enacted in Massachusetts, a state with a population more than 20 times larger than Iceland's, for example, mistakenly characterized in the press as "a sweeping set of genetic privacy protections," illustrates this point. Under this new law, written informed consent is a prerequisite to predictive (but not diagnostic) genetic testing, and to disclosing the results of such tests by entities and practitioners that provide health care.[25] The law also limits the uses that insurers and employers can make of genetic information. However, it places no limitations on how researchers and biotech companies

that engage in projects that require the use of identifiable samples and identifiable genetic information conduct their activities. Apparently those who drafted the statute were under the impression that they need not be concerned about protecting research subjects because research with human subjects is regulated by the federal government, failing to recognize that many activities of genomic companies do not fall under the jurisdiction of the federal regulations.

Policy recommendations

My Boston University colleagues in the Health Law Department and I have argued in the past that a major step to achieving genetic privacy would be the passage of a comprehensive federal Genetic Privacy Act.[26] The primary purpose of this law is to give individuals control over their identifiable DNA samples and the genetic sequence information extracted from them. The model act explicitly provides that individuals have a property interest in their own DNA – and this property

interest gives them control over it. Control could also, however, be obtained by requiring explicit authorization for collection and use, including research and commercial use. In the absence of authorization no one should know more about an individual's genetic makeup than that individual chooses to know, and the individual should also know who else knows (or will know) their private genetic information. Genetic privacy law should:

· Recognize individual genetic rights, particularly:

—the right to determine if and when their identifiable DNA samples are collected, stored or analyzed
—the right to determine who has access to their identifiable DNA samples
—the right of access to their own genetic information
—the right to determine who has access to their genetic information
—the right to all information necessary for informed decision making in regard to the collection, storage

and analysis of their DNA samples and the disclosure of their private genetic information

· Limit parental rights to authorize the collection, storage, or analysis of a child's identifiable DNA sample so as to preserve the child's future autonomy and genetic privacy

· Prohibit unauthorized uses of individually identifiable DNA samples, except for some uses in solving crimes, determining paternity or identifying bodily remains

· Prohibit disclosures of genetic information without the individual's explicit authorization

·Strictly enforce laws and institutional policies

· Provide accessible remedies for individuals whose rights are violated

· Institute sufficient penalties to deter and punish violations

Current U.S. state laws at best offer some economic protections, and a patchwork of genetic privacy protections. But existing state laws have significant gaps and inconsistently regulate those who engage in DNA banking and genetic research. Nevertheless, existing privacy laws provide models and a foundation that can be built upon to protect genetic privacy and empower individuals in this genomic era. Until comprehensive federal legislation is passed, U.S. citizens will have to rely on those who create and maintain DNA banks to design, implement and enforce self imposed rules to protect individuals.

DNA contains uniquely personal, powerful and sensitive information about individuals and their families. Some individuals want to know as much of this information about themselves as possible, and may be willing to share this information with their families and beyond. Others would rather remain ignorant about their own genetic makeup, and thus

their risks for future illnesses, or at least want to keep others ignorant of their genetic makeup. Individual choices are best served by policies and laws that place primary control over an individual's DNA and genetic information in the hands of the individuals themselves.

---

1. Portions of this chapter are adapted from P. A. Roche and G. J. Annas, "Protecting Genetic Privacy," *Nature Reviews Genetics* 2 (2001).

2. See, e.g., Cal. Health & Saf. Code sec. 120980 (West 2000); Conn. Gen. Stat. sec. 19a-583 (West 1999); and Fla. Stat. sec. 394.4615 (West 2000).

3. See, e.g., Committee for a Study on Promoting Access to Scientific and Technical Data for the Public Interest National Research Council, *A Question of Balance: Private Rights and Public Interest in Scientific and Technical Databases* (Washington, D.C.: National Academy Press, 1999).

4. Department of Health and Human Services, "Standards for Privacy of Individually Identifiable Health Information, Final Rule," *Federal Register* 65 (2000).

5. Thomas Murray, "Genetic Exceptionalism and 'Future Diaries': Is Genetic Information Different from Other Medical Information?," in *Genetic Secrets: Protecting Privacy and Confidentiality in the Genetic Era*, ed. M. Rothstein (New Haven: Yale University Press, 1997).

6. George J. Annas, "Privacy Rules for DNA Databanks: Protecting Coded 'Future Diaries'," *Journal of the American Medical Association* 270 (1993).

7. W. Safir, "Senate Inquiry," *The New York Times*, October 23 1993.

8. Paul R. Billings, Jonathan Beckwith, and Joseph S. Alper, "The Genetic Analysis of Human Behavior: A New Era?," *Social Science & Medicine* 35 (1992).

9. Murray, "Genetic Exceptionalism and 'Future Diaries': Is Genetic Information Different from Other Medical Information?,"

10. E. Marshall, "Which Jefferson Was the Father?," *Science* 283 (1999).

11. Dorothy M. Nelkin and Susan Lindee, *The DNA Mystique: The Gene as Cultural Icon* (New York: W. H. Freeman, 1995).

12. M. Huggins, M. Bloch, and S. Wiggins et al, "Predictive Testing for Huntington Disease in Canada: Adverse Effects and Unexpected Results in Those Receiving a Decreased Risk," *American Journal of Medical Genetics* 42 (1992); A. C. DudokdeWit et al., "Distress In Individuals Facing Predictive DNA Testing for Autosomal Dominant Late-Onset Disorders: Comparing Questionnaire Results with In-Depth Interviews," *American Journal of Medical Genetics* 75 (1998).

13. American Society of Human Genetics and American College of Medical Genetics, "Ethical, Legal and Psychological Implications of Genetic Testing in Children and Adolescents: Points to Consider," *American Journal of Human Genetics* 57 (1995).

14. *Norman-Bloodsaw v. Lawrence Berkeley Laboratory*, 135 F.3d 1260, 1269 (1998).

15. George J. Annas, Leonard H. Glantz, and Patricia A. Roche, *The Genetic Privacy Act and Commentary*

(Health Law Department, Boston University School of Public Health, 1995) (available at www.bumc.bu.edu/sph/Lw); Patricia (Winnie) Roche, Leonard Glantz, and George J. Annas, "The Genetic Privacy Act: A Proposal for National Legislation," *Jurimetrics Journal* 37 (1996).

16. *Moore v. U. California*, 793 P.2d 479, 271 Cal. Rptr. 146 (1990), and see George J. Annas, "Outrageous Fortune: Selling Other People's Cells," in *Standard of Care: The Law of American Bioethics*, ed. George J. Annas (New York: Oxford University Press, 1993).

17. National Bioethics Advisory Commission, "Research Involving Human Biological Materials: vol. 1: Ethical Issues and Policy Guidance," (Rockville, MD: 1999).

18. David Korn, "Genetic Privacy, Medical Information Privacy, and the Use of Human Tissue Specimens in Research," in *Genetic Testing and the Use of Information*, ed. Clarisa Long (Washington, D.C.: American Enterprise Press, 1999).

19. H. T. Greely, "Iceland's Plan for Genomics Research: Facts and Implications," *Jurimetrics Journal*

40 (2000); H. Jonantansson, "Iceland's Health Sector Database: A Significant Head Start in the Search for the Biological Holy Grail Or an Irreversible Error?," *American Journal of Law and Medicine* 26 (2000); George J. Annas, "Rules for Research on Human Genetic Variation—Lessons from Iceland," *New England Journal of Medicine* 342 (2000).

20. E. W. Clayton et al., "Informed Consent for Genetic Research on Stored Tissue Samples," *Journal of the American Medical Association* 274 (1995).

21. George J. Annas, Leonard H. Glantz, and B. F. Katz, *Informed Consent to Human Experimentation: The Subject's Dilemma* (Cambridge, MA: Ballinger, 1977).

22. 45 C.F.R. sec 46. 101 et seq. (1991 revision).

23. A. M. Dershowitz, "Karyotype, Predictability and Culpability," in *Genetics and the Law*, ed. A. Milunsky and George J. Annas (New York: Plenum Press, 1975).

24. J. Cumings and G. R. Simpson, "Bush Readies Plan for Legislation to Prevent Genetic Discrimination," *The Wall Street Journal*, June 25 2001.

25. 2000 Massachusetts Acts Chapter 254; R. Misha, "New Law Gives Genetic Privacy Protection," *Boston Globe*, August 23 2000.

26. See *supra* note 15.