Am. J. Hum. Genet. 57:727–731, 1995

Certificates of Confidentiality: A Valuable Tool for Protecting Genetic Data

Charles L. Earley and Louise C. Strong

Department of Experimental Pediatrics, The University of Texas M. D. Anderson Cancer Center, Houston

Summary

Protecting the confidentiality of genetic research data is an important aspect of genetic research that has been discussed in various forums. Research data must be protected to prevent discrimination and its use in litigation. The certificate of confidentiality was created to protect the subjects of alcohol- and drug-abuse studies, who may be engaging in illegal activities. As revised in 1988, the certificate protects investigators engaging in other kinds of studies from being compelled to reveal information about subjects. Because the certificate protects information that could damage a subject's financial or social standing or employability, it is an appropriate tool to use to maintain the confidentiality of genetic data. The Department of Health and Human Services issues the certificates; the procedure for applying for a certificate of confidentiality is presented.

Introduction

Numerous papers have discussed the need for protecting the confidentiality of genetic data (Capron 1990; Cook-Deegan 1990; Touchette 1990; Andrews 1991; Reilly 1991; Schmidtke 1992; Wertz 1992; Harper 1993; Wulfsberg et al. 1994). This is especially important because most informed-consent forms for data collection state that the information obtained will remain confidential. Investigators would not only lose credibility but would, in effect, breach the contract made with the study participants, if the identity of participants were subject to review by insurance companies or employers. Hence, most investigators should use all available resources to avoid having their data subpoenaed and to prevent having to reveal the identity of participants in research projects to insurance companies or employers.

Theoretically, a mechanism to protect genetic research data has existed for several years, in the form of the certificate of confidentiality, issued by the Department of Health and Human Services. Few geneticists currently utilize this protection. This protection became available in 1988, when the statute concerning certificates of confidentiality was revised to include "biomedical, behavioral, clinical, or other research" (Public Health Service Act 42 USCA 241[d]). Before 1988, the protection provided by these certificates was available only to researchers conducting alcohol- and drug-abuse research. The purpose of the law that established the certificate of confidentiality is to protect the identity of research subjects participating in studies that collect sensitive data about those individuals. The interim guidelines issued by the Department of Health and Human Services state that the certificates will be issued "sparingly" but also state that the type of information being collected will be considered on the basis of the sensitivity of the information. A list of categories of information that the Department of Health and Human Services (1989, p. 3) deems sensitive is provided, including (*a*) "information that if released could reasonably be damaging to an individual's financial standing, employability, or reputation within the community" and (*b*) "information that normally would be recorded in a patient's medical record, and the disclosure of which could reasonably lead to social stigmatization or discrimination." The certificate protects the researcher from being compelled to reveal the identity of a research subject "in any Federal, State, or local civil, criminal, administrative, legislative, or other proceedings" (Department of Health and Human Services 1989, p. 1). This protection was clearly necessary for drug-abuse studies, which collect information on subjects' illegal activities. However, the certificate can be a valuable tool for genetic researchers, whose data can reveal not only information on the individual that might be used for identification, but also information that could, in part, identify others or that could reveal information on relationships affecting others, including nonpaternity (which has a reported frequency of ~1% in the general population (Sasse et al. 1994)) or incest. Additionally, individuals could be financially devastated if, because of findings from their participation in a genetic study, they are denied insurance cover-

Received January 30, 1995; accepted for publication May 23, 1995.

Address for correspondence and reprints: Charles L. Earley, J.D., Department of Experimental Pediatrics, Box 209, The University of Texas M. D. Anderson Cancer Center, 1515 Holcombe Boulevard, Houston, TX 77030.

0002-9297/95/5703-0023$02.00

age or employment. Social stigmatization can result if individuals are identified as carriers of genes that may cause certain conditions. The potential for social stigmatization is demonstrated by the Greek Orthodox Church in Cyprus, which, as an option for individuals whose partners are carriers of the gene that causes thalassemia, permits them to choose a different partner (Modell 1992). Even though it is permitted, only 3.3% of the couples have elected to break off relationships (Angastiniotis 1992). Without special protection, geneticists could find themselves in court, confronted with the choice of either revealing information that they assured subjects would be confidential or facing contempt charges for not revealing data.

The Need for Protection

Clearly, there is a need to protect the identity of research participants and the resulting genetic research data. Instances of discrimination against individuals who have taken part in clinical genetic screening have been reported (Billings et al. 1992). An HMO has even required subsequent prenatal screening in a family in which a child was born with a genetic condition. When the fetus tested positive for the mutation, the company considered withdrawing coverage for the pregnancy or limiting the amount of coverage, although they reversed this policy when challenged. Although these instances occurred in a clinical/patient-care environment, they should be cause for concern among genetic researchers.

Kass (1993) reported that an individual's choices for future employment are limited, at least in part, by the inability to obtain new insurance. Inability to obtain new insurance limits the individual to companies in which either the insurance coverage is automatic or no screening is conducted. Wertz (1992) reports that an individual was not hired by the federal government because of being an unaffected carrier of the gene for Gaucher disease.

Information identifying an individual as a participant in a genetic study also has implications for current and future employment (Billings et al. 1992; Wertz 1992; Kass 1993). Not only can the information obtained in a genetic study have significant consequences for the participant, but the mere participation in a research study can have severe consequences. There is at least one reported case of an individual being denied insurance simply for participating in a genetic study (Brownlee et al. 1994). This individual was not affected and did not have the genetic mutation for the condition being studied. Kass (1993) reports that this type of discrimination has also occurred for a participant in an HIV study. The subject had informed his physician of his participation in a research project, and in his medical record a note was made of his participation. When this individual applied for insurance, the insurance company noticed this note in the medical record. The insurance company requested the information from the investigators, who denied the request, citing their certificate of confidentiality. The insurance company denied coverage because the individual did not provide the insurance company with the information from the study. The insurance company was concerned that the subject had access to information, i.e., his HIV status, that might cause him to seek insurance and that thus might result in adverse selection.

Adverse selection involves individuals who have knowledge that would lead them to believe that they will need insurance and who withhold that information from the insurance company. This gives the person an unfair advantage over the insurance company, since the insurance company is unable to evaluate the application accurately. Although the company could have conducted an HIV screening, it asserted that it could not evaluate the potential risk of an insurance claim. The individual did not know his HIV status, but, because of merely participating in the study, the individual would be required to determine his HIV infection status in order to obtain insurance. Although, because of this note in his medical record, this individual was not protected by the certificate of confidentiality, this case demonstrates the need for protection of research information. Denial of insurance, when due to research findings or even to participation in a research study, not only affects the immediate coverage but can potentially bias other insurance companies as well.

Some worker's compensation statutes provide that, when there is a preexisting condition, damage awards can be reduced to the percentage of disability attributable to the work-related exposure. Wulfsberg et al. (1994) raised the possibility that, in cases in which individuals with a given disease had been exposed to workplace factors known to promote the disease but who also could be shown to carry a genetic predisposition to the disease, the genetic information could be used in determination of the employer's fault and/or compensation. Fortunately, such information on genetic predisposition has not yet been regarded as relevant in litigation, but there is the potential for its future application.

In addition to the potential use or abuse of genetic information arising from a research study, in determining insurability, employability, or the existence of a "preexisting condition," genetic information and/or biological samples could be of great benefit to law enforcement officials, in establishing identity or biological relationship. As discussed by Andrews (1991), it could be argued that previously obtained biological samples could be tested without demonstration of probable

cause—i.e., a reasonable belief that the individual was involved in a crime—because there is no further physical invasion of the individual. However, the investigator could argue that consent for the sample collection limited the use of the sample to that outlined in the consent form and hence prohibited any additional use or distribution of the information or the sample, as not explicitly consented to by the participant. This conflict could arise in any situation in which an inventory of biological specimens is maintained, whether they initially have been collected for medical care or research purposes. The certificate of confidentiality would prevent the samples—or genetic information gleaned from them—from being used for any puprose other then that covered in the informed-consent form, by virtue of the certificate protecting the identity of the individual who provided the materials.

There are other ways in which genetic information about research subjects could be used in legal proceedings; although these instances have occurred in clinical settings, it is possible for them to also occur in a research setting. Billings et al. (1992) report the case of a couple who decided not to have children of their own, after discovering, on the basis of clinical evaluation, that the wife had a 50% risk of developing Huntington disease. They decided that they did not want to risk passing the disease-allele risk to their children and attempted to adopt a child. Their application was denied because of the high risk of a parent developing a fatal disease. The reasoning was that the potential parent might not be around when the adopted child reached adulthood.

Billings et al. (1992) and Wertz (1992) cite an instance in which an insurance company had refused to pay for necessary medical care for children after a prenatal diagnosis of cystic fibrosis when the parents had decided to carry the child to term. The family previously had had a child with cystic fibrosis, and, when the woman became pregnant again, fetal screening was conducted. The fetus tested positive for two copies of a mutation that is associated with cystic fibrosis. When the parents decided to carry the second child to term, the insurance company considered limiting the coverage for the pregnancy or even withdrawing coverage. The couple had to threaten legal action, to resolve the problem.

Other authors have discussed the disclosure of risk information to relatives who are not a part of the research project (Wertz 1992; Andrews 1991; Gillon 1988). These authors argue that contacting these individuals would create an undue burden on the geneticist. Wertz (1992) suggests that Tarasoff v. Regents of the University of California would be the controlling case for a genetic researcher failing to warn a relative at risk for a known genetic condition. In this case, a psychiatrist was held to have a duty to warn a patient's girlfriend that the patient had threatened to harm her. Genetic researchers may not be held to the same standard as clinicians, however. Currently, no courts have held that there is a duty to warn relatives of their genetic risk (Andrews 1991). If a duty to contact relatives is established as the standard of care, there are many more questions that will have to be answered, including how to maintain confidentiality of the original participants, which relatives' need to be contacted, how many relatives must be contacted, how much effort must be used to locate relatives, and how much genetic counseling must be given to the individuals contacted. It is clear that information obtained from genetic studies can devastate individuals financially, can be used either to discriminate against them by denial of employment or insurance, or can be used to gain information for use in criminal or civil proceedings, without obtaining the individuals' consent.

Protection Provided by a Certificate of Confidentiality

The certificate of confidentiality protects investigators from being compelled to reveal identifying information about subjects participating in their research projects. This protection applies to any legal proceeding, whether civil, criminal, or legislative. Although the investigator still must report communicable diseases to appropriate public health agencies when this is required by law, the certificate does release the investigator from other reporting requirements.

The certificate protects only the confidentiality of research data. This is important for geneticists who are also providing patient care to the subjects. The certificate does not protect information that is considered a part of normal (i.e., nonexperimental) patient care, including that available in the medical record. When an investigator is both collecting research data and providing patient care, completely separate records need to be maintained, to protect the confidentiality of the research data. As described previously, even mention of participation in a research study in the medical record can be used to discriminate against an individual; hence, although such a stricture is perhaps contrary to standard medical practice, the medical record should not reflect even the participation of the subject in research. Even with computerized medical records, it is possible to keep genetic research information in a separate file, safeguarded from individuals who are not authorized to access those records.

Webster's Medical Desk Dictionary (1986, p. 417) defines a "medical record" as "a record of a person's illnesses and their treatment" Bruce (1984, p. 7), who uses the term "health record," defines it as a document

"that identifies the patient and the health care and services provided to him or her." Since much genetic research data is not clinical data, and since many of the findings do not have an effect on diagnosis or treatment, they need not be included in the medical record.

Although insurance companies may vary in the stringency with which they scrutinize medical records or utilize research data or participation to determine insurability, one denial may have far-reaching effects on the individual's opportunities elsewhere. There is no confidentiality for the denial of insurance, since that information is sent to the Medical Information Bureau, a national database used by insurance companies (Kass 1993). Although preventing insurance companies from accessing genetic research information may produce adverse selection for insurance, the need for research subjects to participate in genetic studies should be considered. Many individuals might not participate in medical research if they knew it could adversely affect their ability to obtain insurance or employment.

The certificate does not prevent voluntary disclosure of information. Any information about a subject can be released with request or consent of that subject. However, release of information about a family requires the consent of each of the family members, not just an individual. If subjects are minors or are incompetent, their guardians are able to consent to the release of information. The protection provided is permanent and remains in place even after the death of the subject. After a research subject dies, the investigator cannot be compelled, by any legislative or legal proceeding, to release information concerning that individual. This is important because of the lack of independence of genetic and family study data. The data on each individual may have a tremendous impact on the lives of many other family members.

Theoretically, if the investigator believes that it is in the best interest of the subject, the investigator could release the information without the subject's or family's consent. However, if the subject did not want the information released and if the informed-consent form stated that confidentiality of the records would be maintained, the investigator could be sued for breach of contract and for breach of fiduciary duty (Andrews 1991). The breach of contract could seriously affect the investigator's credibility with the subject, other family members, and future potential subjects, as well as with the institutional review board and agencies from which the investigator receives funding. The legal ramifications of breaching either the contract with the subject or a fiduciary duty to the subject would involve a court awarding monetary damages to compensate the subject who is injured by this breach. In many instances the damages would be nominal, but, in instances in which insurance is denied because of the genetic information released and in which expensive medical treatment is subsequently required, damages could be substantial.

Applying for a Certificate of Confidentiality

In 1989 the assistant secretary for health established interim guidelines for the certificate of confidentiality (Department of Health and Human Services 1989). The assistant secretary for health is the only official who can grant protection under a certificate of confidentiality, until permanent policies are developed. Agency heads and staff office directors can request this protection for grantees' or contractors' projects. Investigators interested in applying for the protection provided by a certificate of confidentiality should write or telephone the Office of Health Planning and Evaluation, Public Health Service, 737F Humphrey Building, Department of Health and Human Services, Washington, DC 20201 ([202] 690-7100), for a complete information packet.

Acknowledgments

This study was supported by Public Health Service grants CA34936 and CA38929 from the National Cancer Institute, by the National Institutes of Health, and by the Department of Health and Human Services.

References

Andrews LB (1991) Legal aspects of genetic information. Yale J Biol Med 64:29–40

Angastiniotis M (1992) Management of thalassemia in Cyprus. Birth Defects 28(3): 38–43

Billings PR, Kohn MA, de Cuevas M, Beckwith J, Alper JS, Natowicz MR (1992) Discrimination as a consequence of genetic testing. Am J Hum Genet 50:476–482

Brownlee S, Cook GG, Hardig V (1994) Tinkering with destiny: the promises—and Faustian bargains—of genetic testing. US News World Rep 117(8): 58–67

Bruce JAC (1984) Privacy and confidentiality of health care information. American Hospital, Chicago

Capron AM (1990) Which ills to bear?: reevaluating the "threat" of modern genetics. Emory Law J 39:665–696

Cook-Deegan RM (1990) Social and ethical implications of advances in human genetics. South Med J 83:879–882

Department of Health and Human Services (1989) Research confidentiality protection—certificate of confidentiality—interim guidance. Department of Health and Human Services, Washington, DC

Gillon R (1988) Genetic counseling, confidentiality, and the medical interests of relatives. J Med Ethics 14:171–172

Harper PS (1993) Insurance and genetic testing. Lancet 341:224–227

Kass NE (1993) Participation in pedigree studies and the risk

of impeded access to health insurance. Int Rev Biol 15:7–10

Modell B (1992) Ethical aspects of genetic screening. Ann Med 24:549–555

Reilly P (1991) Rights, privacy and genetic screening. Yale J Biol Med 64:43–45

Sasse G, Muller H, Chakraborty R, Ott J (1994) Estimating the frequency of nonpaternity in Switzerland. Hum Hered 44:337–343

Schmidtke J (1992) Who owns the human genome? ethical and legal aspects. J Pharm Pharmacol 44 Suppl 1:205–210

Touchette N (1990) Letting the genome out of the bottle. J Natl Inst Health Res 2(9): 17–20

Webster's medical desk dictionary (1986) Merriam-Webster, Springfield, MA

Wertz DC (1992) Ethical and legal implications of the new genetics: issues for discussion. Soc Sci Med 35:495–505

Wulfsberg EA, Hoffman DE, Cohen MM (1994) α_1-Antitrypsin deficiency. JAMA 271:217–222