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INVITED EDITORIAL
Anticipating Unfair Uses of Genetic Information

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The Ethical, Legal and Social Issues (ELSI) Program was established by the Human Genome Initiative of the National Institutes of Health (NIH) and the Department of Energy (DOE) to anticipate problems that might result from the development of a host of new genetic tests (Watson 1991; Healy 1992). Although the commonly accepted indication for any genetic test is its potential to benefit the individual, the founders of the ELSI program recognized that a series of social problems, including stigmatization, unfair discrimination, and uninformed decision making, might arise from increased availability of genetic information. The areas that were highlighted for investigation included employment, insurance, privacy, informed consent, and education. By bringing more investigators into the field of medical and nonmedical uses of genetic information, the ELSI program has fostered new research and has increased awareness both for the genetics community and for the public at large about the workings of some of our basic social institutions.

The American Journal of Human Genetics has supported the ELSI program by publishing results of new investigations, analytical reviews, and letters. In so doing, the *Journal* has risen to the challenge of one investigator who noted a paucity of publications in human genetics journals about the social implications of genetic information (Lippman 1991). Among the publications in the *Journal* have been a statement from The American Society of Human Genetics to the U.S. Congress on genetics and privacy (Reilly 1992); an editorial about eugenics and discrimination (Holtzman and Rothstein 1992*a*); a general review about the impact of genetic information on life, disability, and health insurance (Ostrer et al. 1993); two articles analyzing legislation that may affect the use and potential misuse of genetic information (McEwen and Reilly 1992; Natowicz et al. 1992*b*); a survey of state insurance commissioners, concerning genetic testing and life insurance (McEwen et al. 1992); a review of cases in which individuals reported unfair genetic discrimination based on family history, genetic laboratory test result, or manifest genetic disease (Billings et al. 1992*b*); and letters to the editor, challenging both the applicability of the Americans with Disabilities Act (ADA) to employment discrimination based on genetic constitution (Holtzman and Rothstein 1992*b;* Natowicz et al. 1992*a;* Natowicz and Alper 1993) and the definition of unfair genetic discrimination (Billings et al. 1992*a;* Hook 1992*a,* 1992*b;* Lowden 1992; Natowicz et al. 1992*c*). The current articles include a survey of medical directors of life insurance companies (McEwen et al. 1993) and an analysis showing how six hypothetical cases of unfair discrimination might be affected by the public entities and public accommodations titles of the ADA (Alper and Natowicz 1993).

From the various articles that have been published to date in the *Journal,* common themes have emerged, which are explored further in the current articles. Is unwarranted, unfair, or undesirable genetic discrimination a significant phenomenon currently? If so, will the implementation of new genetic tests result in increased potential for genetic discrimination in the future? Are current legislative and regulatory schemes sufficient to prevent undesirable forms of genetic discrimination?

The collection of cases by Billings et al. (1992*b*) demonstrated that unfair genetic discrimination occurs. Insurers claim that these are not typical, that they are

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aberrations limited to a few examples of the sort that are inevitable in a large classification system. This point of view was reinforced by the survey of state insurance commissioners, which showed that regulators have not received many complaints about genetic discrimination (McEwen et al. 1992) and by Lowden's (1992) letter to the editor, reminding that insurers seek to sell coverage, not to deny it.

Although these observations provide a useful point of departure, they by no means assuage the concerns that prompted the line of inquiry. Consumers who experience unfavorable insurance-underwriting decisions may not be aware that they have recourse through state insurance commissioners. The methods used by Billings et al. were not likely to reach or to elicit responses from a very large segment of potentially affected persons. Most important, because performance of predictive genetic tests is not yet widely feasible, focusing primarily on the present does not adequately recognize the ways in which the development of such tests will create new pressures and incentives to acquire and to use the information.

The survey of medical directors shows that a substantial number would be interested in using this type of information if it were available (McEwen et al. 1993). The current efforts of insurance lobbyists strenuously opposing legislative restrictions on insurance uses of genetic testing is a testimony to their interest. One of the most interesting results of this survey was the number of respondents whose companies supported restrictions on the use of medical tests in underwriting. Even these companies or the medical directors who showed less interest in using genetic tests would find it difficult to refrain from using genetic tests in the face of market pressures if some companies did utilize them. Fear of insuring a disproportionate share of persons with disease-associated genotypes would force them to join the trend.

The ravaged health-insurance market is instructive about the dynamics of competition based largely on risk selection and rating strategies (Friedman 1991). Thus far, similar fragmentation has not occurred in the life- and disability-insurance markets (Ostrer et al. 1993). Currently, 90% of applicants receive life insurance at standard rates. Competition may act as a safeguard to mitigate unfair discrimination. For example, some life insurers provide coverage to applicants who have had kidney transplants, whereas others do not.

On the other hand, when there is a positive but inconclusive association of genotype with increased risk, all companies may err on the side of caution and refuse the risk. In any case, the identification of markers with substantially greater accuracy than family history will inevitably make it more feasible to deny coverage to high-risk individuals who could not be isolated by currently available methods of risk assessment. Many medical directors of life-insurance companies have wished that the genie of genetic testing would stay in the bottle, maintaining the current state of ignorance about genetic risk, which is based almost exclusively on family history. On balance, life and disability insurers may decide to be more inclusive than many health insurers have been and thus may avoid the practices of the health-insurance industry that have focused on competition to cover only the best risks.

Other interesting observations have emerged from the studies in the current issue of the *Journal.* In the survey of life-insurance medical directors, it was disclosed that some companies send medical test results to insurance agents, without the consent of the applicant, thereby violating the applicant's right to privacy. Some medical directors indicated that their companies' protocols for confidentiality were not available in writing. Companies may have developed underwriting guidelines for certain genetic conditions, without having adequate supporting actuarial data. Although not presented in this survey, one medical director told us that prior to his arrival his company denied coverage to individuals less than four feet tall, on the mistaken belief that very short individuals have decreased life spans. This policy was overturned by the medical director (author's unpublished data). In the current study, the directors' responses to the individual hypothetical cases were highly variable. Although such variability may be typical in medical underwriting, this suggests that some decisions may be highly subjective and potentially uninformed. In the hypothetical cases, some medical directors, on the basis of genetic history, applied different ratings to applicants who may have been at similar risk for premature death, i.e., the man with the family history of Huntington disease and the woman with the family history of breast cancer (McEwen et al. 1993).

In their current article, Alper and Natowicz (1993) have provided a useful service by hypothesizing the range of situations in which personal genetic information may be used unfairly in areas other than employment and life, health, and disability insurance, including automobile insurance, adoption, dental licensure, qualification for a mortgage, admission to medical school, and health club membership. These authors have attempted to define genetic discrimination as dis-

tinct from other bases of discrimination. In order for those who are subject to such genetic discrimination to be protected by the ADA, however, the definition of disability provided by the act must be determined to include genetic discrimination as such. Under the ADA, disability is defined as "a physical or mental impairment that substantially limits one or more major life activities, a record of such an impairment, or being regarded . . . as having such impairment" (42 U.S.C.A. 12102 [West 1991]). These authors present an imaginative idea for extending the protection of ADA against genetic discrimination, to areas outside employment and insurance, by using the public entities and public accommodations titles (II and III) of the ADA (42 U.S.C.A. 12131–12189 [West 1991]).

Most of the analyses in the *Journal* have argued that current legislative and regulatory schemes are insufficient to prevent undesirable forms of genetic discrimination. Considerable discussion has focused on whether the ADA includes within its scope of protection carriers of autosomal recessive conditions and asymptomatic or presymptomatic individuals whose genotype indicates some association with disease (Holtzman and Rothstein 1992*a,* 1992*b;* McEwen and Reilly 1992; Natowicz et al. 1992*a,* 1992*b*). Particular attention has been directed to whether the regulations developed by the Equal Employment Opportunity Commission (EEOC) (titles I and V) and the U.S. Department of Justice (titles II and III) to interpret the ADA include such individuals within their definition of protected disability. Before discussing the substantive merits of this question, clarification of the legal status of such regulations is in order.

As Natowicz et al. (1992*b,* p. 470) have pointed out, regulatory agencies "cannot lawfully change the substance of the laws they interpret." They go on to assert that courts, rather than the EEOC, will have the final word on whether the ADA prohibits genetic discrimination. Although this assertion may be true in some sense, it overlooks an important feature of administrative law, thereby leaving a potentially misleading and perhaps unintended impression that the EEOC's regulatory interpretation carries little weight and that courts will decide these matters independently of the regulatory decisions made by such agencies as the EEOC. A well-settled principle of administrative law makes it likely that courts confronted with this question will defer to the agency's regulatory definition as long as that definition is not arbitrary and capricious and does not conflict with the express intent of Congress. As the U.S. Supreme Court stated in 1984 in Chevron v. Natural Resources Defense Council, "if . . . the court determines Congress has not directly addressed the precise question at issue, the court does not simply impose its own construction on the statute, as would be necessary in the absence of an administrative interpretation" (467 U.S. 837 [1984]). As long as Congress has not expressly addressed the "precise question at issue," the agency interpretation must be upheld, so long as it is reasonable. Such agency interpretations of the ADA therefore carry a presumptive legal weight.

Those who seek protection, in the ADA, from genetic discrimination may be simultaneously encouraged and dismayed by the interpretations embodied in regulations by the several agencies charged with implementation of the ADA. At least two statements from the EEOC have raised substantial doubt about ADA protection against genetic discrimination, if not a presumption that carriers and nonsymptomatic individuals are not included in the ADA definition of physical or mental impairment (Blumenthal 1991; Thornton 1991). Unfortunately, these two EEOC statements and commentators' ensuing discussion of them focus primarily on the first prong of the definition of disability as a physical or mental impairment (Fed. Reg. 35740–35741 [July 26, 1991]). Persons at risk of genetic discrimination, especially those who are asymptomatic or only mildly affected, may prefer not to be covered under this first prong, since, in order to be included under it, they must argue that their genotype is a physical or mental impairment. Such a conclusion is precisely what they want to avoid, especially if they can be protected under another aspect of the ADA definition.

Even if the type of genetic discrimination defined by Natowicz et al. falls outside the scope of protection of the first prong of the ADA definition, carriers and nonsymptomatic individuals with "abnormal" genotypes are most likely to be covered by the third prong of the definition (Fed. Reg. 35742–35743 and 35549–35550 [July 26, 1991]). The regulations of both the EEOC and the Department of Justice state that an individual who does not satisfy either of the first two prongs may be protected by the third prong, which provides that a person perceived by an employer or public accommodation to have an impairment that substantially limits a major life activity is protected from discrimination. This protection includes persons who have no impairment at all but who are erroneously believed by their employer or other covered entity to have a substantially limiting impairment. As an example, the regulations state that, in the case of an employer who discharged an employee on the basis of a totally unfounded rumor

that the individual was HIV infected, the employer's perception of the individual as disabled constitutes prohibited discrimination under the ADA, *even though the individual has no impairment at all.* As the regulations state, "an individual rejected from a job because of the 'myths, fears, and stereotypes' associated with disabilities would be covered under this part of the definition of disability, whether or not the employer's or other covered entity's perception were shared by others in the field and whether or not the individual's actual physical disability would be considered a disability under the first or second parts of this definition" (Fed. Reg. 35743 [July 26, 1991]).

Although agency regulations interpreting the scope of ADA protection appear to cover individuals at risk of genetic discrimination, disturbing aspects of the agencies' interpretations remain a cause of concern, particularly since, in reviewing agency regulatory interpretations, courts are likely to follow the doctrine of judicial deference. The ADA does not limit employment entrance medical examinations and inquiries to job-related health information. In a 1991 letter to the NIH-DOE Joint Subcommittee on the Human Genome, the EEOC Deputy Counsel stated that nothing in the statute or its legislative history suggests that Congress intended to limit employer-sponsored medical tests to job-related conditions and that it would therefore be beyond the scope of the agency's regulatory authority to limit genetic tests to job-related ones (Thornton 1991). This is especially problematic in light of the fact that the agency has added to its regulations for such medical information a confidentiality exception that is neither in the statute nor in the legislative history. Among the conditions that the statute requires for employer medical examinations and tests are that such tests must be administered to all employees and that the results must remain confidential. The statute expressly allows exceptions to this confidentiality provision, for supervisors or first-aid and safety personnel who need to know in order to facilitate an individual's health and safety on the job (42 U.S.C. §12112 [c]). The EEOC regulatory guidelines add a confidentiality exception for insurers, with no basis in the legislative history of the discussions of test confidentiality (Fed. Reg. 35739–35752 [July 26, 1991]). Presumably, this confidentiality exception for insurers was added to facilitate the underwriting of insurance benefits by allowing access to health risk information collected from the employer-sponsored medical tests. Title V of the ADA exempts insurance underwriting from the act's discrimination prohibitions, so long as the employer does not use increased insurance costs as a reason for a dismissal or refusal to hire (42 U.S.C. 12201 §501). Even the legislative history discussing this exemption for insurance underwriting, however, does not mention a confidentiality exception to allow insurers or self-insured employers access to entrance medical examination results. The EEOC addition of such an exception arguably exceeds the agency's scope of authority.

Alper and Natowicz have recognized that the ADA may offer protection against genetic discrimination, beyond the contexts of insurance and employment. They make plausible arguments that Titles II and III of the ADA should cover instances of genetic discrimination against such persons as a prospective adoptive parent or a dental license applicant. The regulatory interpretations of these titles insure that public facilities and testing procedures do not constitute barriers to persons with disabilities. For example, the regulation applicable to professional licensing examinations requires test procedures to accommodate persons with disabilities (Fed. Reg. 35572 [July 26, 1991]). The regulations also prohibit public entities from "administering a licensing or certification program . . . that subjects qualified persons with disabilities to discrimination on the basis of disability" (Fed. Reg. 35718 [July 26, 1991]). These authors have highlighted scenarios that fall within the plain meaning of the statute's language but that apparently have not been anticipated by the agency regulations. These regulations should be reassessed and expanded to include instances of discrimination of the sort described in this article.

The ADA thus provides substantial but incomplete protection against genetic discrimination in employment. It provides virtually no protection against genetic discrimination in insurance or self-insured employee benefits. Title V expressly states that conventional insurance underwriting does not constitute prohibited discrimination. The statute clearly states that this provision does not alter existing state insurance regulations or Employment Retirement Income Security Act (ERISA) exemption of self-insured entities. The EEOC regulations place limits on underwriting discretion by requiring that differential treatment of disability be based on increased risk (Fed. Reg. 35753 [July 26, 1991]). Although there is a basis for this limit in the legislative history of the ADA, the provision effectively violates the McCarran-Ferguson Act, in which Congress expressly left insurance regulation to the states (except for antitrust authority) (15 U.S.C.A. §1013 [a]-[b] [West 1985]). The requirement of the EEOC for sound actuarial data would add an obligation not required by

more than half of the states' insurance statutes on underwriting. Thus, despite support in the legislative history of Congressional committee deliberations, this requirement is unlikely to be upheld, because of both its conflict with the McCarran-Ferguson Act and the acknowledgment by the ADA that state laws control insurance underwriting. It is striking that the ADA and the Human Genome Initiative were debated during the late 1980s and approved in the same year (1990), yet ADA does not clearly recognize or explicitly address the potential for genetic discrimination that has been analyzed by the investigators published in the *Journal.*

Because the ADA does not provide remedies for those who have perceived themselves to be victims of unfair discrimination in insurance, what other recourse might be available? Previously we noted that ERISA (29 U.S.C. §1001–1381 [West 1985]) does not prevent genetic discrimination in self-insured employer health benefits. Although all states have general statutes prohibiting unfair discrimination between individuals of the same risk class (i.e., equal expectation of morbidity or mortality) for life, health, and disability income insurers, such statutes have not been tested for unfair genetic discrimination (McEwen et al. 1992; Ostrer et al. 1993). Some states have augmented these statutes to provide protection for individuals with certain genotypes, but generally these latter are restricted to a limited number of conditions. Where these statutes have been litigated for other unfair-discrimination claims, courts have frequently held that individuals have no private cause of action; hence, successful litigation of such a claim would require action by the state insurance commissioner.

To fill this regulatory gap, some state legislatures have grappled with drafting new laws to restrict the use of genetic information in the underwriting process (McEwen et al. 1992; Ostrer et al. 1993). These statutes are inherently difficult to draft, because the language may be either too restrictive, thereby not providing coverage for many genetic conditions, or so broad that medical underwriting for insurance may be eliminated altogether.

The fact that problems of unfair genetic discrimination must be analyzed by projecting into the foreseeable future necessarily entails some uncertainty and discomfort when public-policy responses are being recommended. Such uncertainty should not be a reason to postpone all consideration or even prophylactic action. Both the difficulty and genius of the ELSI program are to prevent problems before they happen. Gaps exist in the ADA and with state laws, which, if uncorrected, could result in unfair discrimination; however, hastily crafted legislative responses may create unintended consequences. These observations argue for more careful and informed measures, rather than for no action at all.

The ELSI program has brought the practices of many institutions into scrutiny by a host of new investigators. The intent of the program is not to bash private employers or insurers or to draw lines in the sand. The potential exists for unfair or fraudulent use of genetic information by individuals and by those who interact with them. New remedies must balance the concerns of all affected parties, to prevent unintended consequences, such as widespread unfair genetic discrimination or market failures in insurance. Identifying all of the genes in the human genome and the phenotypic effects that are associated with mutation will alter how we view ourselves and how we manage our health. Unlike the eugenics movements of the early 20th century, genetic discrimination against individuals will be fueled by empirical knowledge rather than by inference of genotypes. By anticipating possibilities for discrimination, with an air of open inquiry, we will be able to develop policies that assure fairness and justice to the members of our society.

References

Alper JS, Natowicz MR (1993) Genetic discrimination and the public entities and public accommodations titles of the Americans with Disabilities Act. Am J Hum Genet 53:26–32

Billings PR, Alper JS, Beckwith J, Barash CI, Natowicz MR (1992*a*) Reply to Hook and Lowden: the definition and implications of genetic discrimination. Am J Hum Genet 51:903–905

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

Blumenthal R (1991) Letter from Acting Director of Communications and Legislative Affairs, EEOC, to Representative Bob Wise, Chairman, House Subcommittee on Government Information, Justice, and Agriculture, Washington, DC, November 22

Friedman E (1991) The uninsured: from dilemma to crisis. JAMA 265:2491–2495

Healy B (1992) Hearing on the uses and misuses of genetic information. Hum Gene Ther 3:51–56

Holtzman NA, Rothstein MA (1992*a*) Eugenics and genetic discrimination. Am J Hum Genet 50:457–459

Americans ——— (1992*b*) Reply to Natowicz et al. Am J Hum Genet 51:897

Hook EB (1992*a*) Genetic distinctions are not necessarily examples of genetic discrimination. Am J Hum Genet 51: 897–898

——— (1992*b*) Muddling genetic discrimination. Am J Hum Genet 51:899–901

Lippman A (1991) Research studies in applied human genetics: a quantitative analysis and critical review of recent literature. Am J Med Genet 41:105–111

Lowden JA (1992) Genetic discrimination and insurance underwriting. Am J Hum Genet 51:901–903

McEwen JE, McCarty K, Reilly PR (1992) A survey of state insurance commissioners concerning genetic testing and life insurance. Am J Hum Genet 51:785–792

——— (1993) A survey of medical directors of life insurance companies concerning use of genetic information. Am J Hum Genet 53:33–45

McEwen JE, Reilly PR (1992) State legislative efforts to regulate use and potential misuse of genetic information. Am J Hum Genet 51:637–647

Natowicz MR, Alper JK, Alper JS (1993) Genetic conditions and the scope of the Americans with Disabilities Act. Am J Hum Genet 53 (in press)

Natowicz MR, Alper JK, Alper JS (1992*a*) Genetic discrimination and the Americans with Disabilities Act. Am J Hum Genet 51:895–897

——— (1992*b*) Genetic discrimination and the law. Am J Hum Genet 50:465–475

——— (1992*c*) Reply to Hook: genetic discrimination and public safety. Am J Hum Genet 51:898–899

Ostrer H, Allen W, Crandall LA, Moseley RE, Dewar MA, Nye D, McCrary SV, et al (1993) Insurance and genetic testing: where are we now? Am J Hum Genet 52:565–577

Reilly P (1992) ASHG statement of genetics and privacy: testimony to United States Congress. Am J Hum Genet 50:640–642

Thornton EM (1991) Letter from Deputy Counsel, EEOC, to Paul Berg and Sheldon Wolff, NIH-DOE Joint Subcommittee on the Human Genome Project, August 2

Watson JD (1991) The human genome initiative: a statement of need. Hosp Prac (Off Ed) 26:69–73