

4.1.3 Third-Party Access to Genetic Information

The rapid pace of development and dissemination of genetic testing has made it possible to generate information about individuals across a wide and growing spectrum of genetic variations associated with disease risk. The prospect of access to and use of such information by third parties who have a stake in an individual's health raises ethical concerns about confidentiality and potentially inappropriate use of genetic information.

Patients who undergo genetic testing have a right to have their information kept in confidence, and a variety of state and federal laws prohibit discrimination by employers, insurers, and other third parties based on genetic information they obtain about an individual.

Physicians who provide and interpret genetic tests, or who maintain patient records that include the findings of genetic tests, have professional ethical obligations to:

- (a) Maintain the confidentiality of the patient's health information, including genetic information.
- (b) Release a patient's genetic information to third parties only with the patient's informed consent.
- (c) Decline to participate in genetic testing at the request of third parties (for example, for purposes of establishing health care or other benefits or coverage for the individual) except when at the patient's request and with their informed consent.

AMA Principles of Medical Ethics: IV

Background report(s):

CEJA 3-A-16 Modernized *Code of Medical Ethics*

CEJA Report E-A-93 Physician participation in genetic testing by insurance companies

CEJA Report E-A-91 Genetic testing by employers

CEJA Report A-A-91 Ethical issues in carrier screening of cystic fibrosis and other genetic disorders

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INTRODUCTION

Traditionally, insurance companies have been free to require medical testing before issuing health insurance policies. Insurers have also inquired about an applicant's previous test results, including routine biochemical profiles, urinalyses, electrocardiograms, and x-rays. Insurers have relied on medical testing primarily for two reasons: (1) to “classify” policy holders according to their risk of illness when determining policy premiums;^{2(p.47)} and (2) to avoid “adverse selection” by obtaining the same information about an applicant's medical condition that the applicant may already have. (Adverse selection occurs when individuals apply for insurance in greater amounts because they know they have a higher risk of disease or death.^{2(p.5)})

As a result of the “human genome project,”³ the ability to test people for their risk of genetic disease may increase dramatically. Research from the project is expected to identify genes that contribute to the development of coronary artery disease, cancers, and other illnesses.⁴ Health insurance companies may employ new tests that identify genetic risks of disease, just as they have used existing genetic tests as well as other information that predicts a person's risk of requiring medical treatment. Previous experience with genetic testing suggests that abuses may occur. In some cases, insurers have discriminated unfairly on the basis of genetic test results. In one study, researchers solicited reports of possible genetic discrimination through advertisements to professionals in clinical genetics and related fields. Twenty-nine respondents reported multiple instances of genetic discrimination by insurers. There were threats to deny coverage to a family because a fetus tested positive for cystic fibrosis⁵ and repeated refusals to issue an individual insurance because of hereditary hemochromatosis, despite the absence of any symptoms of the disease. In a survey of 794 members of the National Society of Genetic Counselors and the International Society of Nurses in Genetics by the Office of Technology Assessment, 68 cases were reported in which a patient may have experienced difficulties in obtaining or retaining health insurance because of genetic testing.^{6(pp.20X-20I)} The human genome project will enable physicians to detect an increasing number of genetic predispositions for disease and may lead to further discrimination by insurers in the absence of societal policy and controls concerning the use of genetic information.

The use of genetic testing to deny applications for health insurance would aggravate the gaps in access to health care. In 1987, about 10% of the nation's population was uninsured for the entire year; another 10% was uninsured for part of the year.⁷ In response to this problem, the American Medical Association has approved universal health care coverage for all Americans as a national goal.⁸ However, if insurers were to deny health insurance to applicants because of an elevated risk of genetic disease, health care will become less accessible to many more Americans.

Denial of health insurance on the basis of genetic testing is problematic also because of its effects on patient willingness to undergo such testing. If a positive test result would compromise the patient's access to insurance, the patient may not agree to particular tests that are necessary for an adequate medical evaluation. In this informational report, the Council will address the use of genetic testing by health insurance companies to identify those at risk for developing certain diseases. Genetic testing by health insurers will involve the participation of physicians. This report was prepared to help physicians assess when their participation in genetic testing by insurers is appropriate and does not result in unwarranted discrimination against individuals with genetic abnormalities.

MEDICAL TESTING TAT HEALTH INSURANCE POLICIES

Applicable Underwriting Principles

Insurance operates by spreading risk over a number of people so that many individuals who could have a loss, but do not, help pay for the losses of the few who do sustain losses.^{2(p.4)} Private insurance operates on the principle that the cost of insurance (premiums) should be proportional to the risk that a loss will occur.^{2(p.4)} “Risk classification,” in turn, is a process by which an individual's premiums may be determined.^{9(p.354)} The goal of risk classification is to determine the applicant's expected losses and to place insureds with similar expected losses into the same class so that each may be charged the same rate.^{10(p.1069)} Thus, there is an inherent tension between the principles of risk spreading and risk classification; as risk classification becomes more refined, less risk spreading occurs.

The most important underwriting factors used to classify insureds are age, current health status, and future health status.^{2(p.6),11(pp.783-84)} Current health status is assessed from the application, the Attending Physician's Statement, a physical examination and medical testing. Future health status typically is predicted on the basis of current or past health conditions and the potential for exacerbations or recurrences.^{2(p.6)} Underwriting based on genetic testing raises concern because genetic tests do not measure the extent of disease. Rather, genetic tests indicate the risk that an asymptomatic applicant will develop disease at some future date.

Genetic testing by health insurers primarily affects the market for individual policies. Group insurance and individual insurance are fundamentally different products, and their underwriting practices differ substantially. Medical underwriting is limited generally to the individual and small-group insurance markets.^{2(pp.4243)} Large-group insureds, on the other hand, are issued policies without direct consideration of the insurability of individual members of the group. Large groups are “experience-rated;” their premiums, as well as their access to insurance, are based on actual claims paid within the prior insurance period to members of the group.^{2(p.5)} Approximately 90% of the privately insured population is covered by group policies;^{2,12} roughly 10 to 15% of people with health insurance are subject to medical underwriting.^{6(p.193)}

The use of medical testing primarily in individual insurance is important for two reasons. First, because individual insurance comprises a small share of the total health insurance market, genetic testing should not be necessary to preserve financial stability in the insurance industry. A substantial majority of health insurance is purchased on a group basis, without regard to individual health records. The industry is unlikely to suffer serious financial losses if the use of genetic tests is limited or prohibited.^{13(p.303)} As discussed later in the report, financial hardship should not be a problem even for companies that issue only individual or small-group policies. Second, when genetic testing is used only individual or small-group markets, the people who are tested suffer discrimination simply because they do not have the opportunity to participate in large-group policies through work. Such discrimination is unfair because the availability of adequate health care is a basic right⁴ that should be allocated according to principles of equity and need, not in terms of the employment a person happens to have.

Use of Genetic Testing

Currently, health insurers use genetic information obtained through personal or family medical histories, but there are apparently no insurers that require genetic testing.^{6(p.197)} How new genetic tests will be used by health insurers is necessarily speculative, but there are some data that address the question. In a 1991 survey of private, certain third-party payors (commercial insurers,

Blue Cross/Blue Shield plans, and health maintenance organizations), the Office of Technology Assessment asked the payors about their views on genetic testing.^{6(pp.194-202)} When asked about the future, the insurers responded that it was unlikely that genetic testing would be required in the next five to ten years. On the other hand, the majority considered it likely that they would use genetic information from medical histories in their underwriting in the next five to ten years.^{6(pp.197-199)}

These results are consistent with an earlier survey by the Office of Technology Assessment that examined general medical testing practices by individual and small-group health insurers. The survey revealed that fewer than 4% of applicants are asked to undergo physical exams or blood/urine tests during the underwriting process.^{2(p.9)} Information about test results and disease predisposition is more commonly acquired by insurers from Attending Physician's Statements (APS), which health insurers request from 10 to 85% of their applicants (depending upon type of insurer-commercial, Blue Cross Blue Shield or HMO).^{2(p.9)} The APS responses submitted by physicians typically include the patient/applicant's medical records after the patient authorizes release. The survey results led the Office of Technology Assessment to conclude that the introduction of new tests into the underwriting process will occur not because of direct testing by health insurers, but because of diagnostic testing by the medical profession and the availability of medical records to insurers.^{2(pp.21-22)}

Other factors may cause more direct genetic testing by insurers in the future, however. The human genome project will present insurers with access to more accurate and less expensive methods to screen persons for genetic predisposition for disease. Biotechnology companies now target insurers as "important" utilizers of genetic testing in the future and plan their marketing strategies accordingly.^{15(p.248)}

Competitive forces also may drive insurers to engage in extensive genetic testing. If some insurers begin to make increasingly more sophisticated medical predictions, other companies will be pressured to utilize the same technology.^{16(p.117)}

Justifications for Testing

Adverse Selection

"Adverse selection" is technically defined as the tendency of persons with knowledge of their poorer than average health expectations to apply for or renew insurance to a greater extent than persons with average health risks.^{12(p.1817)} Concerns about adverse selection are two-fold: (1) low-risk insureds will unfairly subsidize high-risk insured; and (2) through inadequate rates and large insurance purchases by high-risk individuals, the insurers' financial solvency is jeopardized.^{10(p.1063)} By collecting medical information about its applicants, insurers can avoid losses from applicants who have undergone medical testing and withheld testing results.¹⁷

Justifying the use of genetic testing with concerns about adverse selection is questionable. First, the argument is overinclusive. In essence, adverse selection is an information imbalance between insurer and insured.^{10(p.1063)} With respect to genetic testing, for example, an imbalance will exist only if the insured has been tested prior to applying for insurance and withholds the results from the insurer. If the insured has not been tested, and many people at risk never get tested, there can be no imbalance of information.

Second, insurance companies are able to offer group plans without relying on medical testing to prevent adverse selection. If costs can be predicted and premiums calculated for large groups

without medical testing, the same should be possible for small groups and individual policies. In many states, health insurers have “open enrollment” periods (e.g., during one month of the year) in which individual plans are available without the usual efforts to measure the applicant's risk of illness. In such cases, policies are assigned a standard “community” rating that is analogous to the common rating of large-group plans. In its recommendations for health insurance market reform, the American Medical Association has identified community rating as a critical element to assuring the Association's goal of universal coverage.⁸

Third, adverse selection is a legitimate concern only if the medical testing at issue reveals information that is truly useful in predicting morbidity and mortality. However, genetic testing generally would have low predictive value. Many genetic diseases, such as Duchenne's muscular dystrophy, cystic fibrosis and Huntington's disease, are rare and do not contribute significantly to morbidity and mortality in the population as a whole.^{2(p.19)} Diseases with a genetic component contribute more significantly to morbidity and mortality and therefore are of greater interest to insurers. These diseases, such as cancer, heart disease, diabetes and emphysema, have some genetic component, but the disease is multifactorial and the onset of the disease, severity of symptoms and efficacy of treatment depend upon the interaction of environmental as well as genetic factors.^{2(p.132),16(p.114)}

Genetic tests are poor predictors of disease and even poorer predictors of disabling disease. Genes are often characterized by “incomplete penetrance”-- many individuals who carry the gene never will show manifestations of the disease, as with retinoblastoma.^{2(pp.139-140),16} When a gene becomes manifest, it will be characterized by “variable expression”--the extent of the gene's effects may differ widely from person to person.⁴ Among individuals with sickle cell anemia, some die within the first years of life, while others survive into their fifties.¹⁸ In many cases, behavioral modification can limit the impact of a genetic risk of disease. Exercise and dietary restrictions can improve the health of individuals with diabetes or coronary artery disease. Even in cases in which a genetic abnormality will cause serious disease, the effects may be delayed for many years. People who develop Alzheimer's disease remain asymptomatic until at least their 50's or 60's. Because of their poor predictive value, genetic tests would have a high false positive rate and would result in many individuals being denied insurance unfairly.

The inherent limitations in the accuracy and reliability of any clinical laboratory test will increase the possibility of false positive genetic tests. For example, when a test is made more “sensitive” so that fewer positive specimens will be missed, it will also be less “specific” and identify more negative specimens as positive.^{2(p.23)} Test results may also vary among individual laboratories performing the tests because not all laboratories have similarly stringent quality controls in place to ensure accuracy.^{2(p.24)}

In sum, the use of genetic testing cannot be justified by concerns about adverse selection. In many cases in which testing would be employed, the applicant would not be aware of his/her health risk. Further, the tests often are not sufficiently predictive of disease to provide material information.

Use of genetic testing to avoid adverse selection would be justifiable only if the information it reveals is essential to the underwriting process. However, just as insurers adjust their premiums in large group plans to take into account the possibility of adverse selection, they can do so with small-group or individual policies. The premiums for all policyholders can be increased to reflect the possibility that some of the policyholders will have an elevated genetic risk for disease. Because of the low predictive value of genetic tests, the increase in premiums for all policyholders would likely be relatively small. Consequently, there is little reason to worry that

low-risk individuals will consider health insurance too expensive if they have to subsidize those at higher risk of genetically influenced diseases.

Fair Discrimination

Some experts argue that restrictions on the types of risk classifiers, including genetic testing, would constitute “unfair discrimination” under the Unfair Trade Practices Act (UTPA), a model law promulgated by the National Association of Insurance Commissioners and adopted in some form in all fifty states.^{19(p.718)} According to this view, the UTPA imposes an affirmative duty on insurers to segregate all insureds with health risks from the pool of insureds without those risks,^{12(p.1810)} and the UTPA would therefore require insurers to classify all statistically relevant differences between groups of insureds.^{11(pp.783-786)} Increasingly sophisticated and refined classification, it is argued, is necessary to avoid subsidization of high-risk insureds by low-risk insureds and to remain competitive in the industry.^{11(pp.783-786)} Under this theory, both diagnostic and predictive testing is required by the UTPA. However, while the UTPA permits classification according to statistical associations with loss, it does not require such classifications. The UTPA only requires that insurance companies treat individuals at similar risk similarly.^{9(pp.381-382),20,21}

Restricting genetic testing is not necessarily unfair to the low-risk policyholders. In group plans, individuals pay the same premiums, and would not be subject to genetic testing, even though the result is that low-risk individuals would “subsidize” the premiums of high-risk individuals. Moreover, if high-risk individuals were denied insurance, much of the costs of their health care would have to be assumed by the government which, in turn, would pass on those costs to low-risk individuals through the tax system.

Fairness to insureds does not demand that all statistical differences in loss be classified. Rather, fairness to insureds requires a balancing of statistical significance and public policy. Too strict a concept of fair discrimination ignores the public policy favoring individual health insurance. The health insurance industry, unlike most other industries, is a private system with a social purpose.” Society has chosen private insurance as the primary vehicle for spreading risks of illness and death. Because of the societal importance of private insurance, insurers enjoy low income tax rates on premiums (about 2%). Employer-based insurance plans also are given favorable tax treatment to encourage employers to provide such benefits.^{22(p.80),9(p.398)} It is unfair for the industry to enjoy a preferred status, but also be permitted to “dump” higher risk insureds because they represent a decline in profit. In its recommendations for health insurance market reform, the American Medical Association has identified community rating, guaranteed renewability and the elimination of preexisting conditions clauses as critical elements of the Association's policy of ensuring universal health care coverage.⁸

In other types of insurance, certain risk classifications are prohibited because of broader policy concerns. For example, property insurers may be restricted in their use of geographic location when rating homeowners insurance (redlining) because of the discriminatory effects of the rating practiced. An important concern is that redlining tends to have disproportionate effects on minorities and exacerbates the socioeconomic disadvantages that they already suffer. Similarly, with many genetic tests, the impact of their use will fall disproportionately on the racial or ethnic groups that have a higher incidence of the genetic abnormalities being tested for.²⁴

Raising insurance rates because of abnormal genetic tests is problematic also because individuals are penalized for traits over which they have no control.²⁴ Often, insurance rates are raised to encourage responsible behavior. Thus, for example, smokers pay higher rates than nonsmokers; individuals with high cholesterol levels may also pay higher rates. In these cases, there is a

genetic component to the higher risk, but there is also an element of personal responsibility, and the higher rates are designed to influence personal behavior. With genetic tests, however, there is nothing the individual can do to affect the outcome.

The insurance industry has resisted legislative proposals for national health insurance, insisting that it is in a better position than the government to manage health care costs.^{25(p.1805)} Unless a public financing system were to become available, then, private insurers should accommodate those persons who most need insurance to pay for their medical care. Any undue burden that would result for insurers could be alleviated by government subsidies. The insurance industry is in a far better position than high-risk individuals to lobby the government for adequate health care subsidies.

It also would be more efficient economically to require private carriers to insure people at risk of developing genetically influenced disease than to permit genetic testing to exclude the higher risks from the private insurance market. Health care costs may be lower if the higher risk individuals receive care through the private health care system. Uninsured people are more likely to defer preventive or early care and end up having to receive more expensive interventions later.

Economic Forces

It has been argued that competition within the industry to attract the most desirable risks will force more genetic testing.^{11(p.789),26(p.57)} If one company begins using such tests, it would be able to offer lower rates to individuals who do not have a specific predisposition for disease. Individuals offered lower rates are more likely to purchase insurance from the company that conducts testing, leaving the higher risk individuals for the nontesting companies.^{26(p.58)} The argument is flawed. If no company uses genetic testing, either because of self-imposed restraint or legislative prohibition, competitive forces will not drive insurers to use genetic tests as part of the underwriting process.²⁷

CONCLUSIONS

Access to health care and health insurance is a fundamental need of all individuals. In its recommendations for health insurance market reform, the American Medical Association has identified community rating, guaranteed renewability and the elimination of preexisting conditions clauses as critical elements to assuring the Association's goal of universal coverage. Consistent with these critical elements, people should not be denied health insurance, or have their premiums raised, because they have an elevated genetic risk for developing disease. Physicians therefore should not participate in genetic testing by health insurance companies to predict a person's predisposition for disease. As a corollary, it may be necessary to maintain separate files for genetic testing results to ensure that the results are not sent to health insurance companies when requests for copies of patient medical records are fulfilled. Physicians who withhold testing results should inform insurance companies that, when medical records are sent, genetic testing results are not included. This disclosure should occur with all patients, not just those who have undergone genetic testing. The Council recognizes that competitive pressures affect the ability of health insurance companies to forgo medical testing. Consequently, regulations or legislation may be needed to preclude the use of either genetic testing or genetic test results. Without such regulations or legislation, a company might be deterred from voluntarily forgoing genetic testing because of concerns about financial disadvantage with respect to competitors.

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CEJA Report E – A-91 Genetic Testing by Employers

INTRODUCTION

Over the next 15 years, under the auspices of the federal government's "human genome project", scientists will try to map in detail each of the human cell's estimated 100,000 genes.¹ The knowledge derived from the project will enable physicians to detect an increasing number of diseases and predispositions for disease. It is expected that researchers will identify genes that contribute to the development of Alzheimer's disease, alcoholism, coronary artery disease, the different cancers and virtually every other illness.² In addition to enhancing the ability of physicians to diagnose disease, the knowledge from the genome project will result in better preventive and therapeutic measures.

Potential applications of information gained from the human genome project extend well beyond the setting of medical care. Employers, insurers and law enforcement agencies all will have uses for genetic testing techniques. In many cases, these uses will provide important social benefits. DNA fingerprinting can establish with greater certainty the identity of a criminal; it can also exonerate the innocent defendant. However, our past experiences with genetic and other medical testing suggest that abuses may occur. Some companies may have restricted employment opportunities of individuals who carry the sickle cell trait³ even though there was no scientific basis for the restrictions.⁴ In addition, employment discrimination has occurred repeatedly against individuals because of their medical problems. Previously, irrational fears led employers to deny jobs to patients with cancer or epilepsy.⁵ Individuals with HIV infection continue to be victims of employment discrimination.⁶

In this report, the Council will address the use of genetic testing by employers to identify employees (or potential employees) who are at risk for developing certain diseases. Genetic testing by employers will involve the participation of physicians. This report will propose guidelines to help physicians assess when their participation in genetic testing by employers is appropriate and does not result in unwarranted discrimination against individuals with genetic abnormalities. The Council's guidelines are summarized in an opinion at the conclusion of the report.

WORKPLACE TESTING

Employers will have a number of potential justifications for genetic testing in the workplace. In some cases, there may be an argument in favor of testing for public health reasons. Companies have expressed concern about the possibility of an employee's genetic susceptibility to illness from exposure to a chemical or other substance in the workplace. In addition, employers may not want to hire individuals with certain genetic risks for jobs that bear on the public's safety. Other justifications are based not on concerns about health but on concerns about costs, specifically the costs to the *company* of hiring workers with a genetic risk of disease. Individuals who have a heightened risk for certain illnesses may be less attractive as employees; on average, they may be able to spend fewer years in the work force, and they may impose greater health care costs on the employer.

Since the acceptability of genetic testing turns on the purposes for which the testing is proposed, each of the proposed justifications for testing will be considered separately.

- 1. Future Unemployability.** Employers may be reluctant to hire individuals who have a genetic predisposition for developing a disabling illness like cancer or coronary artery disease because these individuals may become prematurely unable to work. By excluding those at risk for becoming unable to work, employers may be able to lower their costs of recruitment and training.

As an ethical matter, however, future unemployability is not an adequate basis for performing genetic tests. Genetic tests are poor predictors of disease and even poorer predictors of disabling disease. Genes are often characterized by "incomplete penetrance"; that is, many individuals who carry the gene will never show manifestations of the gene. When the gene becomes manifest, it will be characterized by "variable expression" - the extent of the gene's effects may differ widely from person to person. Among individuals with sickle cell anemia, some die within the first years of life while others survive into their 50s.⁷ In many cases, behavioral modification can limit gene expression. Patients at risk for diabetes can modify their diet, as can patients at risk for coronary artery disease. Even in cases in which the gene will ultimately cause a disabling disease, the effects of the gene may not appear for some time. For example, the onset of Huntington's disease does not occur until the patient is between the ages of 30 and 50 years.^{8(p.163)} Consequently, the use of genetic tests would result in individuals being denied employment well before they became unable to work. In sum, genetic tests would have a high false positive rate and therefore result in many individuals being denied employment unfairly.

Exclusion on the basis of future unemployability is problematic also because it can seriously undermine the principle that underlies the protection of disabled individuals from employment discrimination: disabled individuals should not be denied employment when their disability does not interfere with their ability to perform a job. If individuals could be denied jobs because of future inability to perform, then anyone with an HIV infection could be denied employment as could individuals with other diseases that lead to premature loss of the ability to function in the workplace.

Legal rules are in accordance with this ethical analysis. When construing statutes that forbid employment discrimination against the disabled, courts have consistently rejected an employer's argument that it should be able to deny employment to applicants whose future work might be compromised by health problems.^{9,10(pp.124.126)} While these statutes do not apply to a large percentage of the workforce, the recently enacted Americans with Disabilities Act will prohibit virtually all employers with 15 or more employees from discriminating on the basis of disability.^{8(p.114)} Under the Disabilities Act, medical testing will not be allowed unless the testing is related to the applicant's actual ability to perform the job.¹¹

Employers should not be forbidden entirely from considering a person's future qualifications for a job, and the Disabilities Act will probably permit some consideration of future employability. An individual who would not be able to continue employment for more than a very short period of time, whether for health or other reasons, need not be treated the same as someone who can work on a long-term basis.¹² Important factors in assessing future employability are the likelihood that the employee would no longer be able to work and the length of time before the ability to work would be lost. However, employers can make valid distinctions on the basis of an individual's health status without using genetic tests. People who will become unable to work in a short period of time can be identified by medical testing that measures the effects of genes rather than the genes themselves.

2. Increased Health Care Costs. Employers may not want to hire individuals with a predisposition for cancer, Alzheimer's disease or other illnesses because these individuals might impose higher health care costs on the employer.

Many of the considerations that counsel against genetic testing to assess future employability apply here as well. Because of incomplete penetrance, variable expression and delayed manifestation, genetic tests have poor predictive value also when used as a method for limiting health care costs. In addition, protection against discrimination on the basis of disability would be vitiated if health care costs could be used as a criterion of employment. Individuals with disabilities typically have higher than average health care bills. Consequently, the Americans with Disabilities Act does not recognize higher health care costs as a basis for screening potential employees.⁹ The Act does permit employers to take health risks into account when issuing employee health and other insurance.¹³ However, the Act expressly prohibits

employers from using risk underwriting for insurance as a subterfuge to evade the anti-discrimination purposes of the Act.¹³

From the perspective of society's economic interests, denying employment on the basis of higher health care costs rarely makes sense. Whether or not the person with a genetic abnormality is employed, society will face the same health care costs. (An important exception would occur when a person has a genetic susceptibility to injury from exposure to a chemical in the workplace, a situation that is discussed separately below.) If the person is denied employment, however, there can be no countervailing benefit from the person's ability to contribute productively in the workforce. Thus, if the disabled are working, society's economic interests are better served, as are principles of equity and justice.

There also would not be any unfairness to employers. Since all employers would have the same ethical obligations, any increases in costs would apply across the board. However, if some employers end up with a disproportionate burden of health care costs, it would be appropriate for the government to assist in the form of high risk insurance pools, tax credits or other subsidies.

3. Public Safety. In some cases, employers may want to use genetic tests to protect the public's safety. For example, employers of physicians or airline pilots may want to test for the gene that contributes to the development of Alzheimer's disease when such a test exists.

While ensuring the health of employees whose work bears on public safety is an important responsibility of employers, genetic testing is not an appropriate tool for meeting that responsibility. As when used for other purposes, genetic tests will have poor predictive value when used to identify workers who might pose risks to public safety. Incomplete penetrance, variable expression and delayed manifestation are problems here too. Thus, most individuals who would be excluded by genetic testing never would present a heightened safety risk. Anti-discrimination law has recognized that individuals might be wrongly denied jobs because of speculative safety risks. Consequently, employers must show that there is a significant or reasonable likelihood of harm to others from having a person with a genetic risk of disease employed before the person can be excluded from the workplace.^{5(p.287),14}

Genetic tests are not only generally inaccurate when used for public safety purposes, they are also unnecessary. A more effective approach to protect the public's safety would be routine testing of a worker's actual capacity to function in a job that is safety sensitive. Airline captains, for example, currently undergo physical examinations every six months. Companies that employ bus drivers or ship operators have begun to use simple neurobehavioral testing on a frequent basis to test for impairment by drugs and other causes.¹⁵ Routine functional testing could be used to detect those who become incapacitated by a genetic disease as the disease becomes manifest. In addition, the testing would detect those whose incapacity would not be detected by genetic tests, either because of a false negative test or because the incapacity was caused by something other than the disease being tested for.

Functional testing might also be required by the Americans with Disabilities Act. According to the Act, an individual cannot be excluded from the workplace on grounds of safety if "reasonable accommodations" by the employer would eliminate the safety risk.¹⁴ If functional testing would be more precise than genetic testing at identifying workers who pose a safety risk, then functional testing would likely be viewed as a reasonable accommodation for the employer.⁹

4. Susceptibility to Workplace Exposures. Since at least the 1960s,¹⁶ there has been interest in screening workers for genetic susceptibility to injury from chemicals or other substances in the workplace. Some occupational health experts have argued that genetic tests can be used to identify workers who are particularly at risk for injury from workplace toxins.¹⁷ In fact, black employees have been screened for the presence of sickle cell trait because of concern that exposure to nitro or amino

compounds would result in sickling of the blood cells.^{3(p.320)} Male workers have been screened for the sex-linked genetic abnormality of glucose-6-phosphate dehydrogenase (G6PD) deficiency because of concern that exposure to oxidizing chemicals would precipitate hemolytic anemia.^{3(p.320)} Genetic screening has also been conducted to identify workers with alpha₁-antitrypsin deficiency on the ground that respiratory irritants might cause chronic obstructive lung disease.^{3(p.321)}

While these genetic tests have been used to advise workers of potential risks and for research purposes, they also may have been used inappropriately to exclude affected individuals from the workplace. For instance, the apparent exclusion of workers with sickle cell trait^{3(p.320)} was based on theoretical considerations that had no basis in fact.^{4(p.372)} To date, there is insufficient evidence to justify the use of any existing test for genetic susceptibility as a basis for employment decisions.^{18(p.1106)}

With greater understanding of genetic disease, researchers may develop tests that are more useful for identifying individuals at genetic risk for occupational injury. However, it is doubtful that a positive result on one of those tests will be a sufficient justification for denying employment to the affected person. The poor predictive value of genetic tests is relevant in this context as well. Many individuals with a positive test either will never express the gene, will express the gene mildly or will not express it for a long time. Consequently, many people would be denied employment unfairly.

There is also a serious concern with false negative tests. If companies adopt a policy of excluding "hypersusceptible" individuals from the workplace, they may relax their efforts to eliminate potential toxins from the workplace since the remaining workers will be able to tolerate a higher level of exposure to the toxins. However, because of testing inaccuracies, some affected individuals will not be detected. If they are hired, then they will face an especially elevated risk of injury.¹⁹

Protecting workers from occupational injury can be achieved much more effectively by offering workers the opportunity to be monitored both for their exposure to potential toxins and for adverse health effects from the toxins. When employees are exposed to lead, levels of lead in the workplace are regularly measured to prevent excessive exposures.^{8(p.106)} Similarly, in workplaces where radioactivity is present, the amount of radioactivity that workers are exposed to is routinely monitored to ensure that the employees do not receive an inordinate dose. If workers develop too great an exposure to the toxin, then they should be transferred to a safer job without loss of salary, benefits, seniority, or opportunities for advancement. Under occupational safety and health law, when employees develop excessive blood lead levels from workplace exposure, the employer must provide an alternative job with existing pay and benefits for up to 18 months.^{8(p.106)}

In addition to offering routine monitoring, employers should notify applicants for employment of the occupational risks that they would face in the job and that genetic susceptibilities might increase their risk. The applicants could then have genetic testing performed by their own physicians and decide whether or not to assume the risk of exposure if the test is positive. Although there is insufficient justification for employers to exclude workers with genetic susceptibilities to injury, potential employees should be able to decide that they are unwilling to accept even a very small risk of injury.

It is conceivable that informing applicants and monitoring workers might not be adequate precautions. For instance, while there are no current examples, researchers may discover that a disease develops so rapidly that significant and irreversible injury would occur before monitoring could be effective in preventing the harm. In such a case, there may be a role for testing to identify those who are genetically susceptible to the disease so they can be excluded from the workplace.²⁰

However, before genetic testing could be used for exclusionary purposes, other requirements would have to be met. The employer would have to demonstrate that the genetic tests are highly accurate, with

sufficient sensitivity and specificity to minimize the risk of false negative or false positive results. In addition, there would have to be empirical data demonstrating that the genetic abnormality results in an unusually elevated susceptibility to occupational injury.

Employers would also have to show that it would be too costly to reduce the risk to the susceptible worker by lowering the level of the toxic substance in the workplace. In order to demonstrate undue cost, the employer would have to show that the costs of improving the safety at the workplace are extraordinary relative to other costs of production. Since the alternative to cleaning up the workplace is genetic testing and exclusion, the employer would also have to show that the costs of improving the safety at the workplace are extraordinary relative to the costs of testing potential employees for genetic susceptibility. These requirements would ensure that the costs of using the toxic substance are not placed on a few individuals but on society as a whole. Since society as a whole benefits from the use of the toxin, society as a whole should pay for its use.

Finally, genetic testing should not be performed without the informed consent of the employee or applicant for employment.

Under the Americans with Disabilities Act, if employers use genetic testing to exclude current workers from certain jobs, they may be obligated to offer them alternative employment.²¹ Since genetic testing is ethically permissible only under very limited circumstances, the obligation to provide alternative employment would likely apply to only a small number of workers. Although the obligation does not apply to applicants for employment, the Disabilities Act permits pre-employment genetic tests only in limited circumstances. While an employer may condition an offer of employment on the results of genetic testing that is job related, testing may not be performed until after a conditional offer of employment has been made.²²

EVADING THE PROHIBITIONS ON TESTING

Even if employers do not use genetic testing, they may still be able to discover whether their workers have certain genetic predispositions for disease.²³ Employers will often have access to the medical records of their employees. In some cases, medical records are obtained if there is a question of the employee's ability to resume work after an illness or accident. Although the employer would not need to receive the part of the medical record that includes genetic information, unnecessary information is often disclosed in response to a request for medical records. The patient's genetic information may also be disclosed if the patient receives treatment related to a genetic condition and files a claim for health insurance benefits. Measures will have to be developed to protect the confidentiality of a patient's genetic status.

For the reasons described in this report, the Council on Ethical and Judicial Affairs has developed the following opinion:

Opinion 2.131: Genetic Testing by Employers

As a result of the human genome project, physicians will be able to identify a greater number of genetic risks of disease. Among the potential uses of the tests that detect these risks will be screening of potential workers by employers. Employers may want to exclude workers with certain genetic risks from the workplace because these workers may become disabled prematurely, impose higher health care costs, or pose a risk to public safety. In addition, exposure to certain substances in the workplace may increase the likelihood that a disease will develop in the worker with a genetic risk for the disease.

1. It would generally be inappropriate to exclude workers with genetic risks of disease from the workplace because of their risk. Genetic tests alone do not have sufficient predictive value to be relied upon as a basis for excluding workers. Consequently, use of the tests would result in unfair discrimination against individuals who have positive test results. In addition, there are other ways for employers to serve their legitimate interests. Tests of a worker's actual capacity to meet the demands of the job can be used to ensure future employability and protect the public's safety. Routine monitoring of a worker's exposure can be used to protect workers who have a genetic susceptibility to injury from a substance in the workplace. In addition, employees should be advised of the risks of injury to which they are being exposed.
2. There may be a very limited role for genetic testing in the exclusion from the workplace of workers who have a genetic susceptibility to occupational illness. At a minimum, several conditions would have to be met:
 - a. The disease develops so rapidly that serious and irreversible illness would occur before monitoring of either the worker's exposure to the toxic substance or the worker's health status could be effective in preventing the harm.
 - b. The genetic testing is highly accurate with sufficient sensitivity and specificity to minimize the risk of false negative and false positive test results.
 - c. Empirical data demonstrate that the genetic abnormality results in an unusually elevated susceptibility to occupational illness.
 - d. It would require undue cost to protect susceptible employees by lowering the level of the toxic substance in the workplace. The costs of lowering the level of the substance must be extraordinary relative to the employer's other costs of making the product for which the toxic substance is used. Since genetic testing with exclusion of susceptible employees is an alternative to cleaning up the workplace. The costs of lowering the level of the substance must also be extraordinary relative to the costs of using genetic testing.
 - e. Testing must not be performed without the informed consent of the employee or applicant for employment.

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INTRODUCTION

The recent development of a prospective screening test for the cystic fibrosis gene has raised concerns about the benefits and risks of carrier screening. This report examines the ethical principles governing carrier screening for genetic disorders and their application to carrier screening for cystic fibrosis. The scientific and economic issues of carrier screening for cystic fibrosis are addressed in a companion report by the AMA's Council on Scientific Affairs.

Ethical Principles Implicated by Carrier Screening for Genetic Disorders

Beneficence. The principle of beneficence requires that the physician promote the health and welfare of patients. It also includes working to prevent or remove harm from patients. One of the primary purposes of carrier screening is to reduce the pain and suffering and relieve burdens produced by genetic disorders. Ethical considerations are raised most often in those cases where the disorder is of a serious nature but no curative therapy exists. Reduction of pain and suffering is achieved primarily through the reduction of incidence of the disorder, i.e., by avoiding the births of affected children. This may be accomplished through the use of artificial reproductive methods such as artificial insemination or egg donation or by performing prenatal diagnosis followed by selective abortion. Alternatively, knowledge of carrier status allows couples who would attempt pregnancy in any case the opportunity to prepare for the possible birth of an affected child.

Patient Autonomy. The principle of patient autonomy requires that patients are given the opportunity to make their own determinations regarding medical treatments. Therefore, patients must give informed consent before genetic testing is performed.

An important purpose of carrier screening is to maximize autonomous decisionmaking for individuals with regard to reproduction. Previously, couples ordinarily only discovered that they were both carriers of a genetic disorder after the birth of an affected child or because close relatives manifested the disorder. Now, ideally, preconception screening can determine the status of individuals who have a high probability of being carriers, as well as those who may not have had previous indication that they may be carriers.⁸⁻²⁹ For example, when an individual has a close relative affected by a genetic disorder, the individual and his or her spouse can now be tested; in the past, many such couples may have chosen to forgo childbearing rather than risk the birth of an affected child. Knowledge that neither or only one partner in a couple is a carrier may relieve the couple of some anxiety about reproduction. Couples in which both are carriers may choose from several reproductive options, including use of artificial reproduction, avoidance of conception, and use of prenatal diagnosis to detect and, if appropriate, terminate an affected fetus.

Justice/Fairness. The principle of justice requires that both benefits and burdens of medical technology be distributed equitably. Concern has been raised regarding issues of access to and cost of screening and counseling services. Currently, access to prenatal care, particularly to genetic counseling services, is skewed in favor of well-educated couples from the middle and upper-income levels.¹⁴ Problems with access to medical services is not confined to obstetrical and genetic services. However, because genetic risk factors for genetic disorders are generally uniform across socioeconomic boundaries, the ethical principle of justice dictates that access to genetic screening must be uniform across socioeconomic boundaries.¹⁵

Nonmaleficence. Physicians have an ethical duty to avoid harm to patients. Genetic technology frequently raises concerns regarding the communication and comprehension of risks. The principle of informed consent requires that patients be given enough information to enable them to make intelligent choices about their treatments. In the context of genetic screening, harm may arise from the inappropriate communication of results. For both carrier screening and prenatal genetic testing, diagnoses are often heavily dependent on complex estimations of risk which may not be easily understood by laypeople. The communication of complex risk information may be liable to confusion or unnecessary anxiety among patients. The duty of nonmaleficence requires that physicians minimize confusion and anxiety by locating the most effective methods for communicating complex risk calculations and explaining complicated genetic information.

Confidentiality. The principle of confidentiality obligates the physician to maintain the privacy of the patient to the greatest possible degree. Exceptions to confidentiality are made only for situations which present overriding social considerations. The obligation of confidentiality is especially compelling in the context of genetic technology because of the highly personal nature of genetic information. Genetic information about an individual that is revealed to third parties may negatively impact an individual's social relations, employment status, or insurability. Also, decisions which may result from genetic information may implicate intensely personal moral or religious beliefs and values of individuals.

Screening for Cystic Fibrosis

The ability to screen for genetic disorders is not new. Tests for carriers status of certain genetic disorders, such as Tay-Sachs disease and sickle-cell anemia, have existed for some time. In addition, certain types of genetic analysis, such as "linkage analysis,"²⁹ have already been successfully employed to identify genetic disorders in individuals.

Screening for cystic fibrosis has special significance because cystic fibrosis is a relatively common genetic disease. Cystic fibrosis is a potentially lethal autosomal recessive disorder characterized by chronic lung disease and pancreatic insufficiency,^{1,2,29} and is the most common serious genetic disorder among the United States Caucasian population. Approximately one in 25 people of European heritage are carriers for cystic fibrosis.¹ Assuming random mating, roughly one in 400 Caucasian couples will be a two carrier couple.³ A couple in which both individuals are carriers for cystic fibrosis will have a one in four chance of having a child affected by cystic fibrosis. Cystic fibrosis is relatively rare in non-Caucasian populations.^{3,4} Studies have also shown that the frequency of cystic fibrosis is lower among certain white sub-groups. Presently, median survival of a person with cystic fibrosis is 25-27 years, with some individuals reaching age 40 or greater.²

Until recently, direct detection of carrier status for cystic fibrosis was not possible.^{4,5} Within the last few years, however, researchers have isolated a specific deletion ($\Delta F508$) on chromosome seven which is responsible for approximately 75% of cystic fibrosis chromosomes in white individuals in the United States.^{4,5} Other deletions have been identified which are responsible for an additional 10% of cystic fibrosis chromosomes.³¹ In other words, the test has a false negative rate-of at least 15%. Once the false negative rate has been taken into account, testing for this deletion can identify approximately 72% of couples who are at a one in four risk of having a child with cystic fibrosis.^{1,31}

The resolution of the ethical issues raised by carrier screening for CF will have widespread and significant ramifications. Past carrier screening efforts were aimed at genetic disorders which were prevalent only in certain small, well-defined racial or ethnic groups, for instance, screening

for sickle cell anemia in blacks or Tay-Sachs disease in Ashkenazic Jews.¹⁶ Unlike previous screening programs for carriers, testing for CF cannot be limited to a relatively small, well-defined population which is at an elevated risk.³ Due to the absence of significant indications for screening besides European heritage, essentially the entire white population of the U.S. is “at risk” for being a CF carrier.³

This fact has far-reaching social and economic implications. Introduction of carrier screening to the general U.S. population could precipitate a demand for genetic screening and counseling of unprecedented proportions.³ Certainly, current resources, particularly those of the necessary skilled personnel, are not sufficient to meet this demand.¹ In addition, the commercial potential of widespread carrier screening is enormous. The pressure of market demand could influence the course of screening for CF carriers in profound ways.^{1,3}

Screening for cystic fibrosis is also of critical significance because it could be used as the test case for the handling of other genetic disorders. The recent commitment by the National Institutes of Health to map the human genome means that the discovery of the source of many other genetic disorders is imminent. The success or failure of carrier screening for CF may not only impact the course of future carrier detection programs, it also has the potential to shape the role that biotechnology will play in our society.

ETHICAL ISSUES RAISED BY CARRIER SCREENING

Experience with Past Carrier Screening Programs

Previous carrier screening programs have had varying rates of success. Both the successes and failures of past programs can be looked to in order to ascertain minimum requirements for implementing future programs.²⁹

Carrier screening for Tay-Sachs disease, which was begun in the early 1970s, is widely regarded as successful. Several elements of the Tay-Sachs screening program contribute to its success. Tay-Sachs disease occurs among Ashkenazic Jews, a relatively small and well-defined population. The test for Tay-Sachs disease is simple, accurate, and relatively inexpensive. The screening program was preceded by extensive efforts to educate a clearly defined target population of the need for and nature of carrier screening. Pilot tests were also preceded by careful planning which included the active involvement of community leaders and institutions.³ The active involvement of the targeted communities has resulted in a high percentage of participation in screening by the high-risk population.

Screening for Tay-Sachs disease is also successful because of the severe nature of the disease. Tay-Sachs disease is an incurable, progressively degenerative disease which is fatal at an early age. The severity of the disease was a compelling justification for active efforts to prevent births of Tay-Sachs children. Prenatal diagnosis for Tay-Sachs disease was available at the time of the first carrier screening programs, which maximized the reproductive options for two-carrier couples. In addition to avoiding conception or using artificial means of reproduction, a couple could conceive and abort any fetuses afflicted with Tay-Sachs disease.

Screening for sickle-cell anemia was conducted differently from screening for Tay-Sachs disease and yielded negative results. Sickle-cell anemia manifests itself among black populations, also a relatively small and well-defined population in the United States. Unlike Tay-Sachs disease, which is consistently severe and debilitating, sickle-cell anemia can manifest itself with varying degrees of severity. At the time that carrier screening was initiated, there was no prenatal

diagnosis for the disease and artificial means of reproduction were still experimental, so the only option available to a carrier-carrier couple was to avoid reproduction altogether.

Screening programs for sickle-cell evolved rapidly, with little preliminary education of the targeted population, little or poor planning, and inadequate attempts to involve community leaders or other community members in the screening process.³ The objectives of screening were unclear and target groups for screening were poorly chosen. For instance, screening efforts were often aimed at schoolchildren, who had no use for the information. Many programs used tests which did not differentiate between carrier and affected status. In addition, even where carrier and affected status could be distinguished, the communication of screening results frequently confused or ignored the difference between being a carrier for the sickle-cell gene and having sickle-cell anemia.^{3,16}

To complicate matters, a large number of states implemented mandatory screening programs at this time. Adequate safeguards of confidentiality of testing results were not maintained.¹⁶ There were instances of insurance and employment discrimination based on testing results. Lack of prenatal diagnosis for the disorder, lack of legitimate goals of screening, poor communication of goals, and the variable severity of the disease led many black populations to suspect discriminatory motives behind the implementation of mandatory programs.¹⁶

Lessons learned as a result of these screening efforts are invaluable for the development of screening programs for cystic fibrosis. Any future carrier screening for genetic disorders (1) must be based on clear, well-thought out objectives, (2) must be preceded by extensive education and planning, (3) must include substantial community-based support and co-operation, and (4) must have strict safeguards for confidentiality.

There are critical differences, however, between the nature of cystic fibrosis and the type of genetic diseases for which we have previously screened. For instance, the risk of being a carrier is uniform across many different social, economic, ethnic, cultural, and educational groups. Population screening can not be restricted to a small, well-defined target group at an elevated risk. Cystic fibrosis, similar to sickle-cell anemia, manifests itself with varying degrees of severity in individuals. The need for screening may be less compelling for disorders which vary in the degree of debilitation in individuals. While tests for both sickle-cell anemia and Tay-Sachs disease are simple, accurate, and relatively inexpensive, current tests for cystic fibrosis have significantly high false negative rates and may be costly.¹ In order to implement carrier screening for CF in an effective and just way, pilot studies to establish effective methods of education, counseling, and proper communication of risks to all groups are essential before general population screening can begin.

Potential Threats to Patient Autonomy

Although carrier screening is intended to facilitate autonomous and informed reproductive decision making legitimate concerns have been raised regarding potentially detrimental influences of genetic testing on reproductive decisionmaking.^{3,9,12} For those genetic disorders in which there is no effective therapy, the primary intention of screening is to avoid the birth of affected children. While this may ultimately reduce the incidence of genetic disorders, the very ability to avoid the births of affected children may create coercive pressures upon the decisionmaking process. Couples choosing to forgo testing, or to forgo the termination of pregnancy and bear a child with CF may be subject to blame or stigmatization for making an “irresponsible choice.”³ Some couples may be perceived as responsible for “burdening” society with an affected birth which could have been avoided. Couples may be pressured either to abort

any affected pregnancies or to avoid reproduction altogether. For instance, one insurance company tried to deny health care coverage to an infant with cystic fibrosis on the ground that since prenatal diagnosis had been positive for CF, then the disease constituted a preexisting condition. (The company eventually provided coverage.) Blame or stigmatization for certain reproductive choices may be aimed at women in particular, since the responsibility for carrying a pregnancy to term is often regarded as ultimately belonging to the “Perceptions of a couple's responsibility for Causing” the birth of an affected child may exacerbate the morally troubling nature of decisions about reproduction. In addition, if the incidence of genetic disease is reduced through carrier screening, those remaining individuals who are affected may be subject to increased prejudice or stigmatization .

Reproductive choices are regarded by both medicine and society as intensely private decisions. The appropriate choice, in the context of possibly having a child with a genetic disorder, is often uncertain from an ethical perspective. Frequently, evaluations of risks and benefits are heavily dependent on the moral or religious convictions of the individual or couple. For these reasons, the information regarding the reproductive risks and benefits of genetic screening is best evaluated by those who will bear the risks and receive the benefits. ^{11,12} Care should be taken to avoid placing moral judgments or pressure on parental decisions regarding the use of information derived from genetic testing. The Council has previously recognized this general principle in its *Current Opinion 2.12*, on genetic counseling:

Physicians...may decide that they can engage in genetic counseling and screening but should avoid the imposition of their personal moral values and the substitution of their own moral judgment for that of the prospective parents.¹³

It is important that the process of enhancing reproductive autonomy through genetic technology not have the unintended and opposite effect of restricting reproductive options through negative or coercive pressures on decision making. ³

C. Ethical Considerations in the Communication of Test Results

Medicine has traditionally placed high value on the patient's right to confidentiality in medical and health information. Informed consent is required for disclosure of an adult patient's medical information in almost every situation. Exceptions are legitimately made only in the most extreme cases, such as to avert a serious harm to identifiable third parties, ²⁵ to prevent the spread of serious and contagious diseases, or for other overriding social considerations. ²⁶ This principle should also govern the release of the results of carrier screening for genetic disorders.

The results of individual test results from carrier screening will most likely be of interest to parties other than those individuals who are screened. Among those who may be interested in the results are biologically related third parties, employers ²⁷ and insurers. Both employers and insurers may be interested in information regarding the genetic status of individuals for use in calculations for business decisions. However, these relatively narrow interests do not constitute an overriding social consideration which would outweigh the physician's obligation to protect the confidentiality of the patient. In addition, the potential for inappropriate use of this information by these parties has prompted the President's Commission for the Study of Ethical Problems in Medicine to state:

Because of the potential for misuse as well as unintended social or economic injury, information from genetic testing should be given to people such as insurers or employers only with the explicit informed consent of the person screened. Further, the agencies in

question should develop forms for specific rather than blanket consent, to prevent unnecessary disclosures and to ensure the screen control over access.

The stronger challenge to the patient's right to privacy may come from close relatives of the person who is screened and receives a positive test result. The knowledge of carrier status gained from testing may be able to prevent serious harm to identifiable third parties, such as the progeny of relatives. However, interest in the prevention of harm to identifiable third parties, which may override confidentiality in other narrowly drawn situations,²⁵ does not apply strongly in the case of carrier status. The actions of a carrier cannot "cause" or "spread" the disorder in the manner of a communicable disease. Also, the likelihood of preventing a harm may not be great; the possibility that the relative of the carrier and the relative's partner would both be carriers and then give birth to an affected child are still relatively small. Concerns about informing related third parties are also diminished since relatives of identified carriers can be screened themselves. In general, confidentiality should not be breached to inform related parties of an individual's carrier status.

Physician participation in programs which make access to screening for CF contingent upon the disclosure of results to unrelated third parties such as insurers or employers are ethically problematic. Such prerequisites may burden autonomous reproductive decision making and interfere with the duty of the physician to protect the patient's interests. Many individuals may choose to forgo testing rather than risk the disclosure of results to third parties.

Equally problematic are programs which establish substantial or coercive incentives for the couple to make a particular reproductive decision. While individual physicians cannot be responsible for the business practices of health insurance companies or employers, the physician still has ethical obligations to protect the patient's medical interests to the greatest extent possible. Physicians should minimize the potential for inappropriate influences on patient decision making by outside parties. The ethical principles of beneficence and patient autonomy require that patients give specific informed consent for disclosure to third parties. This consent should inform patients which information may be revealed, to whom it may be revealed, and potential and likely uses of the genetic information. Patients should also be informed whether alternative methods of obtaining the same information exist (e.g., the patient could pay for the test independently or the patient could be directed to a community health center which performs testing).

Potential Difficulties in the Communication of Genetic Information to the General Population

For some genetic disorders, less than perfect rates of sensitivity result in a relatively high false negative rate. In the case of cystic fibrosis, 25% of the deletions responsible for CF chromosomes have not yet been identified. Only 50% of couples who are at one in four risk of having a CF child would be identified. The communication of risks to couples is currently complicated, since a negative result reduces the risk of being a carrier but does not eliminate it. Screening performed with the current false negative rate would risk causing a false sense of security for couples who are mistakenly identified as being at no or low risk for having a cystic fibrosis child. It may also cause difficulties in the communication and understanding of the risk factors involved. However, as additional mutations causing cystic fibrosis are identified, the false negative rate will decrease⁴ and the risks of false negatives will be greatly reduced.

Several authors have also suggested that the potential confusion arising from complicated risk estimations justify postponement of population screening programs for cystic fibrosis until a 90 or 95% sensitivity rate could be achieved.² However, the current false negative rate by itself is not an ethical justification for postponing population screening. Negative results would be highly

problematic for couples, since risks would be reduced but not eliminated, complicating already morally problematic reproductive decisions. Estimations of risk may be complex and difficult to comprehend. However, information which could aid the informed decision making of some couples should not be withheld on the basis that not all couples may understand complex information' or that the decisions are morally troublesome. Rather, it is the responsibility of the medical profession to develop effective methods of communicating risk or other complex medical information to patients and the general public. The exact evaluation of those risks is best made on a case by case basis by the particular couple, based on their own personal values, rather than by physicians individually or medicine as a whole.

Another serious concern is the high rate of false positives which results when a screening test is used to detect a low-prevalence disease. Wilfond and Fost have estimated that, even assuming a high rate of test specificity for carrier status, the false positive rate would approach 50%.¹ In the case of CF, half of the couples who are identified as being at an elevated risk for producing a CF child would have been falsely identified as such. Repeat carrier testing after positive results, the development of confirmatory tests, and subsequent prenatal tests for some couples would alleviate or eliminate the false positive rate. The primary concerns for false positives are unnecessary anxiety about carrier status, risks of additional confirmatory testing, and the possibility that significant reproductive choices will be made based on false information. Some couples, particularly those who do not feel selective abortion is an appropriate option for them, may choose to avoid childbearing or to undergo artificial reproduction rather than chance conceiving an affected fetus. However, to withhold screening from all couples because only half of couples would be properly detected would deprive 50% of at-risk couples from important information for their reproductive decision making.¹⁹

Some experts have stated that population screening should not be initiated until the completion of pilot studies which demonstrate effective procedures for counseling and education, develop comprehensible methods for the communication of risks to all socioeconomic and cultural groups, and minimize potential for social stigmatization.^{1, 3} The difficulties encountered by early attempts at sickle-cell screening and the need to minimize inappropriate influences on reproductive decision making highlight the need for programs which include effective education and counseling, ensure confidentiality, and communicate risks in a clear fashion. This need justifies postponing population screening until appropriate pilot studies are completed.

Physician obligations regarding the provision of carrier testing

The potential market for CF screening tests may prompt developers or manufacturers of the test to appeal to physicians and/or the public in order to encourage wide use of the CF carrier test before adequate pilot testing is completed. Patients may receive information about the availability of such tests either on their own or through advertising or marketing efforts by manufacturers of tests, and may be prompted to request screening. What are physicians' ethical obligations regarding carrier testing before adequate information is available to evaluate the consequences of testing?

If individuals have a close relative with CF, then testing the individual or the individual's partner using the carrier test for the DF508 deletion is appropriate.¹ Testing for carriers among close relatives would be almost 100% informative, since directly testing for the DF508 deletion could be combined with linkage analysis in order to minimize the rate of false negatives for this particular sub-group. This small group is already known to be at an elevated risk, and testing can be considered medically indicated.

A physician's obligation to provide carrier testing for cystic fibrosis in absence of an elevated risk is less clear. Prevailing practice norms among geneticists emphasize patient autonomy and fully informed decision making. A study of the attitudes and practices of U.S. geneticists showed that when faced with an uncertain result from prenatal testing, or when the results of prenatal testing were of disputed importance, the great majority of geneticists endorsed presenting the information to the patient or patients, acknowledging any uncertainty or disagreement about the results, and allowing the couple to decide on their own which course of action is most appropriate for them.⁷³

However, the Council has previously stated that a physician may ethically refuse to perform genetic counseling or testing for personal moral reasons provided that the physician alert the parents that genetic services are available elsewhere.¹³ Also the American Society of Human Genetics has stated:

...while it is recognized that testing of highly motivated individuals in the general population may occur, it is the position of the American Society of Human Genetics that *routine* CF carrier testing of pregnant women and other individuals is NOT yet the standard of care in medical practice. (emphasis in original)³⁰

Thus, it would not be unethical for a physician to refuse to perform or refer for genetic screening for CF before pilot studies establish adequate methods of education and counseling for population screening.

Defining physician's ethical obligations to inform patients about genetic tests however, also implicates concerns about legal obligations and liabilities. Suits against physicians who did not inform patients of the availability of tests which could have prevented specific harm have been allowed in most states.^{3,12,32,34,35}

Ordinarily, physicians who comply with the accepted standard of care in providing patients with information about specific genetic tests fulfill their legal obligation to the patient.^{36,45} However, some legal experts have warned that physicians cannot be guaranteed protection from liability for failing to perform or refer a couple who requests CF carrier testing, even if the couple was not at an elevated risk.^{36,37} It is likely that some highly motivated or informed individuals will specifically request testing.³⁰ Failure to perform or refer for testing in these cases is likely to incur liability.^{32,34,39-41}

Further, failing to offer the test to patients, even in absence of a specific request or in the absence of an elevated risk for CF, may also carry a small risk of liability.^{32,34,36,37,40,42,44} A few courts have specifically rejected the claim that a physician's duty to inform patients of diagnostic tests is limited to those instances in which prospective parents ask questions and request specific care.^{45,46} Physicians are likely to be the sole or primary source of information for prospective parents regarding the genetic health of future children.⁴⁷ Some courts may choose to focus on the fact that the prospective parents would consider the results of the current CF tests, while not conclusive, to be material to their childbearing decisions.^{36,45,47} However, a physician's liability in such cases is likely to be highly dependent upon the practices prevalent in the physician's community⁴⁵ the extent to which patients would have access to facilities which perform quality CF testing, and the extent to which the court would consider the test to be experimental.⁴³

In contrast, routinely offering screening tests to couples before adequate safeguards have been established may also exacerbate present problems with testing, and thereby increase liability risks.^{33,41,46} Offering screening in the absence of established methods of education, counseling, and communication of risk information may result in the inaccurate communication of results, inappropriately influencing already morally problematic decisions.

It is important to note that as a matter of public policy, it would be unwise for courts to allow the threat of liability to prompt physicians to offer genetic tests before being accepted as the standard of care.

GENERAL ETHICAL AND SOCIAL POLICY CONSIDERATIONS

Implementation of Population Screening

Mandatory Screening. Mandatory screening for CF carriers within the general population cannot be ethically justified. Mandatory screening could yield some benefits. Some possible benefits could include equal access to screening through the equal allocation of resources, minimization of burden to society through the reduction of the incidence of CF, and promotion of genetic health. However, mandatory testing violates fundamental notions of reproductive autonomy and informed consent. The decision to be tested for genetic information requires subjective evaluations of social, moral, and cultural values. These evaluations should be made by the couple or individual who would undergo screening. Some couples would choose to undergo childbirth regardless of risk estimations, and other couples may not consider the reproductive alternatives, such as selective abortion or artificial insemination, to be appropriate for them.

There is also reason to believe that well-developed and well-executed voluntary screening programs could produce similar benefits without violating fundamental ethical principles of autonomy and informed consents. Screening programs for Tay-Sachs disease indicate that voluntary programs which maximize cooperation and coordination within the communities and the screened populations can achieve high rates of success.

Targeting the appropriate population for screening. Ordinarily, the populations targeted for screening or testing are those at risk, or at an elevated risk, for a disorder. Within the United States, essentially the entire Caucasian population is at an elevated risk for being a CF carrier. However, carrier screening for genetic disorders should be limited in some ways.

Carrier screening should be at least limited to those who are of reproductive age. The knowledge gained from carrier screening is medically significant only in the context of potential risks to the health of future generations. The social risks of information regarding carrier status, such as possible anxiety, stigmatization or discrimination, outweigh any benefits which may be derived from the communication of risk information to those who cannot have children.²²

The optimal time for possessing the results of carrier testing would be before conception occurs. Screening could thus be routinely offered to all individuals of reproductive age. Routine, nonjudgmental screening of all individuals of reproductive age might also serve an educational function. A large population would receive important information about the nature of genetic disease, and the routine nature of testing may minimize misunderstanding or prejudice toward carriers or those affected. Carrier status might become a routine consideration for individuals, similar to ascertaining Rh status, for reproductive decisions. On the other hand, resources may be wasted by testing many who would have little or no use for the information.

Other types of limitations have been suggested. There have been proposals to limit screening to couples specifically planning to have children. This would minimize demands on resources, but would have too many risks. Many pregnancies are not planned, and many couples who do plan their pregnancies do not seek preconception medical examinations. In these cases, reproductive options for couples who both tested positive would be limited by the fact that conception had

already occurred. There are also proposals to limit screening to married couples or couples planning marriage. However, many married couples do not plan to have children and couples who have children are not necessarily married or planning marriage. Limitations based on marital status or intent to have children would not be appropriate.

Other limitations on the population to which screening is offered would not be appropriate. For instance, early prenatal diagnosis procedures were occasionally denied to couples who did not consider abortion an option if an affected fetus were detected.²³ Consensus has since developed among physicians and geneticists that willingness to undergo abortion should not be a requirement for access to diagnostic services.²³ Even for couples who plan to carry an affected pregnancy to term, prior knowledge of the status of the fetus may be invaluable to the couple's preparation for the child. Similarly, limiting access to carrier screening based on the patient's willingness to use the information in a particular way cannot be justified. Limitations on access to screening would also be inappropriate if based on factors such as socioeconomic status, race, sex, ethnicity, except where such factors also determine risk level.

In the context of the allocation of scarce health care resources, an argument may be made for making screening available only to those who specifically request it. However, such a practice risks violating the ethical principle of justice. Under a request-only system, individuals who are better-educated and have greater access to health care information would be much more likely to request, and consequently receive, screening. Others with an equal risk for being a carrier but lacking certain kinds of education or access to information would be placed at a disadvantage for access to screening services.

While routinely offering testing is appropriate, routinely recommending it is not. Uniform recommendations for genetic screening may also carry potential risks. Since the social influences and consequences of increased reliance on genetic technology for this purpose are mainly still unknown, patients should make the choice to utilize the technology in the context of their own values to the greatest extent possible. Routinely offering but not recommending access to screening allows individuals this possibility without undue influence. One exception would be couples in which one individual has a close relative with cystic fibrosis.

Once appropriate ethical requirements are met, decisions regarding the availability of screening will probably have to be on a societal level, since questions of resource distribution and social policy are heavily implicated. Once the availability of carrier screening has been limited to a particular population, then the most just way of distributing the benefits and risks of testing would be to routinely and uniformly offer it among that population.

Ethical Issues Implicated by Carrier Screening for Genetic Disorders Indicate a Need for Coordinated Social Policy

The Need for Broad-based, Well-Coordinated Social Policy to Direct Screening Efforts. The ethical issues which arise from the implementation of genetic screening indicate a need for well-conceived, well coordinated social policy. Prominent among the ethical requirements for screening is the need to ensure uniform access to screening services among at-risk populations. While physicians play a role in providing access to medical services, facilitating access to health care requires widespread cooperation among many entities within society, including physicians, patients, insurers, employers, government, and regulators.

Genetic screening and other genetic technology have the potential for significant impact on society and our social structure. One of the most widely heralded dangers is that of using genetic

screening to promote an inappropriate eugenic agenda. Overt eugenics, such as government-imposed constraints on marriage and reproduction, are unlikely to be implemented. Most overt constraints would be subject to legal or constitutional challenges. A more likely circumstance would be that the aggregate result of individual choices creates societal and cultural norms which substantially influence or limit the scope of autonomous decision making in regard to the use of genetic technology. Avoidance of negative consequences, such as increased marginalization of individuals who are affected by genetic disorders or socially coercive attitudes toward certain reproductive choices, requires careful attention to possible conflicts or problems incurred by the implementation of screening.

One of the most important aims of social policy in this area, then, is to recognize and avert potentially negative constraints or influences on the use of genetic screening. Overall uses for information obtained from carrier screening may also be influenced by the policies of particular institutions.³ For instance, insurance companies which refused to pay for CF screening unless the parents agreed to accept significantly higher premiums for any affected infants which were born would have a coercive effect on reproductive choices. Employers may feel employees who are carriers present a financial risk in terms of the potential health care costs of any affected offspring who may fall under the employer's health plan. It is important to avoid having the narrow interests of a single or small number of parties dictate overall social policy. Particular institutional interests such as insurance companies or employers should not be allowed to discriminate against carriers of genetic disorders, either overtly or through policies which have the ultimate effect of substantially influencing the content of reproductive decision making.

Carrier Screening Should Not Replace Efforts to Find Curative Treatments for Genetic Disorders

Finally, it is important that the development of carrier screening for genetic disorders does not detract from efforts to find treatments or cures for the disorder.² In the case of cystic fibrosis, roughly 30,000 Americans currently have CF, and more children will continue to be born with CF every day.² It is unlikely that even the most effective screening program would totally eradicate the incidence of CF or other genetic disorders. Despite society's best efforts, screening may remain inaccessible to many groups or individuals. For some individuals, alternatives such as selective abortion or the use of artificial reproduction will not be in accord with personal moral values. Also, random genetic mutations not detectable through population screening will continue to produce a small but significant number of genetic disorders in individuals. Efforts to reduce the pain and suffering of individuals born with genetic disorders must continue to include the search for effective treatments and curative therapies.

It is also important to find curative therapies for a particular genetic disorder because the ability to cure a disease reduces or eliminates the number of morally problematic reproductive decisions which often accompany information gained from screenings. Curative therapy would obscure the need to choose between the termination of pregnancy and the birth of an affected child. Also, the risks of false negatives or positive results would be lessened.

RECOMMENDATIONS

The Council on Ethical and Judicial Affairs recommends:

Regarding the use of carrier screening for cystic fibrosis:

- Pilot programs for carrier screening for cystic fibrosis should be initiated to determine effective methods of education, counseling, and communication of genetic risk information to general carrier screening populations.
- The implementation of mass screening for cystic fibrosis carriers should be delayed until pilot programs have determined methods or levels of education, counseling, and communication of risk information which minimize the potentially negative consequences of carrier testing.
- Initiation of carrier testing for adults with a close relative who is affected by cystic fibrosis is appropriate. However, any testing program for this small group of patients who are at an identifiable elevated risk for being carriers should be carried out only within a coordinated system of genetic services, including confirmation of diagnoses through other genetic technology (i.e., linkage analysis, etc), education and counseling.
- The physician has no ethical obligation to routinely offer testing services or provide information about screening services before pilot studies have adequately established methods of testing which minimize risks and negative consequences of testing. Once a consensus has developed as to the general safety and efficacy of screening programs, then ethical obligations are incurred. Physicians should avoid the imposition of personal moral values or the substitution of personal moral judgment for that of the prospective parents. In addition, physicians should strive to ensure that the process of carrier screening includes all possible safeguards against attitudes, influences, or pressures which may stigmatize particular reproductive choices or which facilitate prejudice or stigma against carriers, parents of children affected with cystic fibrosis, or individuals affected with cystic fibrosis.

Regarding carrier screening generally:

- All carrier testing must be voluntary, and informed consent from screened individuals is required. Confidentiality of results is to be maintained. Results of testing should not be disclosed to third parties without the explicit informed consent of the screened individual. Patients should be informed as to potential uses for the genetic information by third parties, and whether other ways of obtaining the information are available when appropriate.
- Allowing third parties access to the results of carrier testing may inappropriately restrict reproductive decisions and is not acceptable.
- Carrier testing should be available uniformly among the at-risk population being screened. One legitimate exception to this principle is the limitation of carrier testing to individuals of childbearing age. In pursuit of uniform access, physicians should not limit testing only to patients specifically requesting testing. If testing is offered to some patients, it should be offered to all patients within the same risk category.
- The direction of testing for cystic fibrosis and future genetic screening tests should be determined by well thought out and well-coordinated social policy. Third parties including

insurance companies or employers, should not be permitted to discriminate against carriers of genetic disorders through policies which have the ultimate effect of influencing decisions about testing and reproduction.

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