Appendix B

Privacy and Control of Genetic Patient Data

introduction

Some of the same recombinant DNA technology that makes human gene therapy possible will also facilitate the identification of many more individuals with genetic diseases than earlier techniques allowed. This new technology should result in a dramatic increase in the amount of genetic patient data that can be collected, much of which has never been available before.[1] However, the ability to gather potentially large amounts of new genetic data about individuals raises questions about rights of privacy regarding that information, as well as the ability of others to have access to it.

WHAT ARE GENETIC PATIENT DATA?

Genetic patient data refer to information collected about an individual relating to his or her genetic constitution. Information of this sort can include a large number of individual traits, ranging from eye color or blood type to predispositions to or presence of various diseases. Since genes determine many personal characteristics, genetic data may reveal important facts about an individual's physical and intellectual status or potential. One's genetic complement is an involuntary endowment, since the genes are passed on from parents, and genetic characteristics are not generally subject to change.

Policies on access to genetic patient data must balance the benefits deriving from disclosure against the need to preserve individual privacy. The benefits to public health and other priorities often determine that medical information be disclosed. Examples of situations in which medical information is used for public good or prevention of harm include reporting child abuse or other criminal conduct, notifying State officials about the presence of communicable disease that might endanger public health, and use of disease statistics in planning priorities for biomedical research. Patients might be harmed, however, as a consequence of disclosing their genetic data. They might be socially stigmatized, have difficulty finding a mate, encounter barriers to obtaining life and health insurance, or be discriminated against when seeking employment.

Genetic patient data are different from other types of disease- related medical information, in the following ways:

- In contrast to communicable diseases, the public at large is not at risk of contracting genetic disease, since it can be transmitted only to progeny. Because of the genetic transmission of the disease, information about close relatives may reveal information about oneself, and vice versa. Closely related individuals can benefit from this information.
- Because some genetic diseases, such as Huntington disease, colonic polyposis, or polycystic kidney disease, may not be expressed until middle or old age, genetic information in some cases provides a look into the future health of an individual.
- Because of the emotional concern of the patient when learning about a genetic disease in their family against which he/she has no defense.
- Future generations may inherit the disease, and therefore have an interest in it.

Those potentially interested in genetic patient data include the patient, his or her family, insurance companies, employers, health care providers, and the Federal Government.

HOW ARE GENETIC PATIENT DATA COLLECTED?

Genetic patient data are collected about individuals in many ways, but the bulk of specific information on genetic traits derives from two main sources: family histories and genetic tests.[2]

A family history can be relatively easy to collect, and most genetic patient data available to physicians are of this type. A family history is usually obtained by asking the patient questions about the presence of diseases in his or her family that are known to be inherited. Histories can often be supplemented by inquiry among other family members. The importance of genetic factors varies between diseases. Recent data on Alzheimer disease indicate that a significant fraction, at least one-third of cases may be genetic (Breitner, 1984; Folstein, 1981; McKusick, 1983), while other diseases, such as PKU, are always due to genetic defects. Variation in the genetic component among different diseases and even among diseases of the same

[1] The number of cloned human genes is an index of this increase in potential genetic patient data The number of cloned human genes reported at the Gene Mapping Meetings has risen from 22 in 1982 to 132 in 1984 (Skolnick, et al., 1984)

[2] These include a variety of biochemical and genetic tests See app A for further information on genetic testing techniques.

type can be due to several factors, discussed in the overview, such as:

- incomplete penetrance,
- variable expression,
- environmental factors,
- different patterns of inheritance: dominant, recessive, or sex-linked,
- multigene traits, and
- multifactorial traits.

As a result of these factors, genetic patient data collected from family histories can alert individuals to personal health risks and statistical likelihoods, but it generally cannot predict with certainty whether an individual with a family history of cardiovascular disease or cancer, for example, will actually develop those disorders.

With reliable genetic tests, it is sometimes possible to determine the presence of genes that can cause disease, permitting more accurate determination of the probability of expressing symptoms. Genetic testing may be performed as a result of information obtained in the family history, or can, in some cases, be initiated to screen for diseases common in a more general population to which the patient belongs.

Genetic patient data are collected in several different contexts. Family histories are recorded when an individual first visits a physician, and generally when a person buys individual life insurance. Genetic testing is often performed in the context of making personal, medical, or reproductive decisions, and such tests are performed at different times for different reasons (Rowley, 1984). Carrier screening can identify individuals who carry one copy of a deleterious gene so that they may be made aware of the risks of having a child with a genetic disease and make a clearly informed decision about having children. Carrier screening has been performed on groups at high risk of carrying certain genes, such as Blacks and Mediterranean populations, who may have hemoglobin disorders, or Eastern European Jews who may carry Tay-Sachs disease.[3]

Prenatal screening is performed to identify possible genetic defects in the fetus and allow parents to decide whether it should be brought to term or if it might require special care when born (see app. A). Prenatal screening is indicated in several situations, including when the mother is 35 years or older, if a previous child were born with a genetic defect, or if both parents are known carriers of a gene which can be detected by such screening (Milunsky, 1980), Screening at birth can identify newborns who require special care, such as PKU newborns who need a special diet, low in phenylalanine. For this reason, newborn screening for PKU is required by most States.

Genetic screening raises many medical, ethical, legal, and economic questions, such as: 1) Can family members crucial for testing be legally coerced to participate in linkage studies (see ch. 1), 2) Should a person of any age have the right to be tested and informed of test results?, 3) Should spouses or parents be permitted to know this information?, 4) Does a child have the right to genetic information held by his parents?, 5) Should physicians inform at-risk individuals of the availability of testing?, and 6) Can the release of information from genetic testing be withheld in employment and health insurance questionnaires? (Kurlan, 1983). One of the most difficult issues is the use of abortion to prevent genetic disease. Other questions include whether the benefits of genetic screening exceed the costs of the procedure, and if so, whether newborn screening should be made mandatory (President's Commission, 1983). The President's Commission for the Study of Ethical Problems in Medicine and Biomedical and Behavioral Research enunciated five principles for genetic screening with the following recommendations:

1. Confidentiality. "Genetic information should not be given to unrelated third parties . . . ;"
2. Autonomy. "Mandatory genetic screening programs are only justified when voluntary testing proves inadequate to prevent serious harm to the defenseless, such as children, that could be avoided were screening performed;"
3. Knowledge. "Decisions regarding the release of incidental findings (e.g., nonpaternity) or sensitive findings (e.g., diagnosis of an XY female) should begin with the presumption in favor of disclosure . . . ;"
4. Well-being. "Screening programs should not be undertaken until the test has first demonstrated its value in well-conducted, large-scale pilot studies . . . .A full range of prescreening and followup services for the population to be screened should be available before a program is introduced;" and
5. Equity. "Access to screening may take account of the incidence of genetic disease in various racial or ethnic groups within the population without violating the principles of equity, justice, and fairness. "

This paper will not discuss further the issues related to the collection of genetic patient data; rather, it will address issues which arise after the data is collected.

[3]Screening for most genetic disorders is performed on a voluntary basis, although most States require screening of newborns for PKU and some other disorders Five States require such testing under all circumstances, 30 permit denial on the basis of religious convictions, and 9 others permit some other bases for refusal, PKU screening not required in three States (Andrews, 1984d) Some other mandatory screening laws, particularly those that involve screening of adults for potential carrier status, have been repealed because of claims made by the affected groups that they were being singled out and discriminated against (Rowley, 1984)

WHY ARE GENETIC PATIENT DATA IMPORTANT?

Genetic patient data can play an important role in the life of an individual, affecting such diverse areas as:

- **choice of spouse;**
- **psychological health**
- **reproductive decisions, such as**
  -decisions to have children
  -decisions to undergo prenatal screening, and
  -decisions to terminate pregnancy;
- **decisions about personal health risks affected by diet, smoking, and health habits;**
- **decisions about the personal health risks connected with certain jobs; and**
- **decisions concerning financial, insurance, and retirement plans.**

These are among the most personal decisions that an individual makes, and it is therefore important that their privacy be ensured. However, as mentioned above, there are others besides the individual who have an interest in genetic patient data, and their interests must also be considered.

Genetic patient data may also be significant because they have the potential for being misunderstood or misinterpreted by the public. Earlier genetic screening programs to identify carriers of sickle cell disease caused some individuals to be stigmatized because they and others did not understand the difference between the carrier state and the disease state. Some of these individuals were mistakenly treated as 'sickly' children or discriminated against in employment or insurance coverage (Rowley, 1984; President's Commission, 1983). This and other examples highlight the need for greater understanding of genetic conditions before using genetic patient data to direct social policy. As more is discovered about the genetic basis of certain diseases, such as alcoholism, schizophrenia, or complex traits such as intelligence, issues of individual privacy relating to genetic patient data may become even more important than they are today.

Privacy and access

In any discussion of the privacy of health records it is important to consider the tradeoffs between an individual's right to privacy and others' interests in having access to the same information. Privacy and access are two sides of the same coin, and to preserve an individual's right to privacy is to deny others that access. If all genetic patient data were made completely private, society would forego the potential benefits accruing from availability of that information, such as planning national biomedical research priorities and preventing potential harm to relatives, Equally unavailable would be data vital to the determination of paternity and the identification of criminals in court cases. In addition, it would be impossible to conduct research on genetic diseases. The benefits, however, must be weighed against the fact that unrestricted access to genetic patient data would violate the autonomy of individuals to reveal only the personal information of their choice. Two models illustrate the ways in which health records are treated: the physician-patient model and the public health model.

THE PHYSICIAN-PATIENT MODEL

The precedent for confidentiality in the physician-patient relationship was set many years before the Hippocratic oath was written (Walters, 1983), and since that time, physicians have held to an ethical code of privacy in matters relating to patient's records.

Utilitarian Justifications.--One way to consider the privacy of the physician-patient relationship is utilitarian: for the physician to effectively treat the individual there must be trust between them. A patient can only be expected to reveal delicate health issues to the physician if the information is to be held in strict confidence. In daily life, a person can control whether or not to disclose personal information to others. One's private thoughts may be represented by a set of concentric circles, with the outermost circles containing information that a person is willing to give to anyone, such as height or occupation, and the innermost circles containing personal information that is reserved only for those closest to him or her, if anyone. In the medical model, a patient allows a physician to enter an inner circle in order to get help with a medical problem, and the physician therefore owes a duty to the patient to keep the information confidential (Walters, 1983). Certain types of genetic patient data may be considered so proprietary that, "Doctors in whose records this information may reside should hold it extremely confidential and should not keep it in the person's general medical file" (Wexler, 1983).

Patient Rights.--Another approach to the physician-patient relationship is centered on the rights of the individual. These rights become particularly important in considering the difference between collecting a family history and performing genetic tests. A patient has direct control over whether to provide a family history to a physician, while genetic testing can be performed on blood, body fluids, or tissues. This technical ability to collect genetic patient data raises two main concerns. First, the patient does not exercise the same discretionary control over information garnered from biochemical testing as he or she does in relating a family history: the patient merely assents or dissents to undergoing the test. Second, blood or tissue samples collected at other times for other rea-

sons may be tested genetically, without the knowledge of the patient.

Even following the guidelines for informed consent, with the patient agreeing to genetic tests, their technical nature increases the risk that the patient does not fully understand the possible significance of the data. Patients may also fail to anticipate the potential harm disclosure might cause him or her. The consent of the patient is required to remove blood or tissue from his or her body, and also to perform tests, but it is important that the patient be informed of all the tests which are done and that a concern for the privacy of the patient extends to the control of tissues removed from his or her body.

Under normal circumstances, health records are not released to third parties, except with the consent of the patient, so that medical information which exists in the record is still under the control of the patient. Nevertheless, current practices involving information release allow little or no control over withholding parts of data. A patient with a genetic trait or disease is rarely able to release only the parts of his or her record that do not contain that information once a waiver is signed, as those waivers are considered as 'blanket' consent for release of their entire medical record. However, even in instances when the physician-patient relationship can be maintained, there are several cases which supersede it and these can be grouped and called the public health model

THE PUBLIC HEALTH MODEL

A physician's duty to protect the privacy of his patient may be superseded by his duty to prevent harm to others, such as the patient family or society in general. For example, a physician must report the occurrence of cases involving gunshot wounds, battered children, and certain communicable diseases (Green and Capron, 1974; Walters, 1983). Government interest in reporting communicable diseases centers on identifying both the disease and those individuals who are at risk of contracting it, and mobilizing efforts to prevent or treat it. With certain communicable diseases, such as gonorrhea, there is a high risk of danger to significant numbers of people, and government involvement may be a way to reduce the risk. With gunshot wounds, it is possible that the injury occurred as a result of an illegal act that may place others at danger, and so government action may prevent harm to others. This concern for the public well-being often places the physician in a difficult ethical position, having to choose between the privacy interests of his patient and the interests of society. This is especially true in the case of psychiatrists who may have reason to believe a patient may become violent, and they must decide whether their belief justifies reporting the patient to the police (Walters, 1983).

Since there are many different issues involved in the disclosure of information, it is instructive to look at several different cases of the disclosure of information, beginning with the disclosure to the patient, himself.

DISCLOSURE TO THE PATIENT

The doctrine of informed consent, so called, was initially developed to assure a patient's self-determination and right to decide whether to undergo health care procedures. One of the most important arguments for an informed patient is that only with adequate information can an individual make informed decisions concerning his or her health or lifestyle, and genetic information can play an important role in these decisions. Another, recently discovered, and perhaps more compelling argument is that informed consent may actually provide numerous physical and psychological benefits to the patient (Andrews, 1984a).

Studies of elective surgery patients have provided the most notable evidence of the beneficial effects of information disclosure. Patients 'briefed' on the nature of surgical procedures and postoperative sensations exhibited a greater capacity to adjust to postoperative stress, needed less pain medication, and had fewer recovery days in the hospital. In another study of hospital patients, one of the chief reasons for refusing treatment seemed to be the occurrence of unexpected procedures which exacerbated patient uncertainty and aroused patient anger (Appelbaum, 1982).

However, the therapeutic effects of information disclosure are not limited to surgery patients. Patients scheduled for endoscopic examination— where a fiberoptic tube for internal viewing is placed down the esophagus and into the stomach—heard a taped description of the sensations frequently experienced during the procedure and subsequently needed less medication to tolerate the examination than those who did not hear the tape. Similar results indicating the benefits of disclosure have been found in studies involving blood donors, burn treatment, and sigmoidoscopy examinations (Andrews, 1984a).

Disclosure also acts as an informal check and balance system whereby a patient may reject a procedure that is being advocated more for the benefit of the practitioner than the patient. Although generally acting in the patient's best interest when they propose diagnostic procedures and therapies, physicians may be motivated by strong financial and professional considerations that place them in a conflict of interest (Schneyer, 1976).

Another potential benefit of informed consent is that it may enhance the quality of physicians' decisions. By

requiring physicians to provide clear and factual information about the risks and alternatives to a given procedure or therapy, they may recognize and account for their own judgment biases and suggest a more thoroughly considered course of action. Additionally, in the course of the physician describing a procedure, the patient may reveal information pertinent to the treatment choice— information which may result in a different choice of action.

There is no consistent or prescribed amount of information due the patient on a national basis, but there are three measures by which the legal system generally determines the patient's right to decide. One is the Reasonable Physician Standard, whereby the physician follows the standards of the community to determine how much, or whether to disclose anything to the patient. The second is the Reasonable Patient Standard, whereby the patient is informed of any and all information necessary or helpful to a reasonable patient. The third is the Individual Patient Standard, whereby the physician must take into account what he/she knows about the individual patient to determine what should be disclosed. Each of these standards carries different weight with different courts, and despite the widespread acceptance of the doctrine and its continued expansion, the patient's right to informed consent has always been and continues to be a qualified one (Andrews, 1984a).

Courts almost unanimously note several exceptions to the general rule: an emergency situation where the patient is unconscious or otherwise unable to authorize treatment, and serious damage will occur if treatment is not undertaken; where the patient is deemed incompetent to make a decision; where a waiver to informed consent is signed by the patient; and where therapeutic privilege is invoked because disclosure poses such a threat of psychological damage as to be unwise from a medical viewpoint (Andrews, 1984a).

Third party access

Several groups besides the individual would have an interest in genetic information gathered about an individual. For example, family members may wish to be alerted to potential health risks revealed by the genetic data about a close relative Also, insurance companies, employers, and the Federal Government have an interest in access to genetic patient data for various reasons which will be described below. In each case, there is conflict between third party access to information and the individual's right to privacy.

A physician's duty to protect the confidentiality of the patient data can be upheld if certain guidelines are followed when disclosing information to third parties:

- there should be a high probability of harm to others,
- the potential for harm should be deemed serious, such as being irreversible or fatal, and
- there should be reason to believe that the information will prevent harm. (President's Commission, 1983, p. 44).

Reasonable attempts for voluntary consent should be made, since it would not be ethical and may not be legal[4] to disclose information without the consent of the patient, and only the relevant information should be disclosed. These guidelines will be considered in the following situations: disclosure to family members, insurance companies, employers, and the government.

DISCLOSURE TO FAMILY MEMBERS

There are many situations in which genetic data about an individual may affect decisions made by close relatives. Genetic data may be of greatest importance to one's spouse or prospective spouse because it may directly affect the couple's reproductive decisions. The reason for disclosure is to prevent direct harm to the unborn and indirect harm to one's spouse. In many cases, one partner would wish to inform the other about possible genetic risks so that together they may make an informed decision about having children. In other situations, the affected partner may prefer not to inform the other, in order to avoid being identified as the cause of having deformed children or being the reason for not having children at all.

Disclosure to a spouse may indeed prevent harm if the couple decides not to have children at high risk of genetic disease. The reasons supporting disclosure of genetic patient data to a spouse increase with both the severity of a potential genetic disease and the probability of the children inheriting it.

Another reason for disclosure goes beyond reproductive decisions to include the need for the spouse and family to know the genetic condition of the affected person in order to make plans to care for them, both physically and financially. For example, if it were known that the provider of a household would develop polycystic kidney disease or Huntington disease, the family would have to plan for the debilitating effects of the disease, significant medical expenses, and future loss of income.

[4] A physician who discloses medical data to relatives or third parties may be sued for damages resulting from violation of the patient's privacy

Since children receive half their genes from each parent, they also have an interest in the genetic data of their parents. The case for disclosure to children is strong because there may be a significant probability of harm that could be reduced if the children were to take health precautions. In families with colonic polyposis, for example, those with the disease are at high risk of developing colon cancer, and preventive removal of the colon can thwart almost certain death from cancer. Knowledge about colonic polyposis can, therefore, be of extreme importance to those at risk.

Genetic patient data may also be relevant to health care of other relatives. In families that carry the gene for retinoblastoma, for example, children are at high risk of developing potentially fatal eye cancer. Knowledge that a relative has the disease may precipitate more careful scrutiny of cousins and siblings who are also at risk, thus potentially saving lives.

The case for access to more distant relatives is generally not as strong as for the immediate family, since the predictive value is lower, but here, too, genetic patient data might alert the person to potential health risks. If the severity of the disease and the degree of risk is high and action can be taken to prevent harm, then disclosure to more distant relatives maybe justified,

Finally, genetic patient data can be of use to children and other relatives of parents affected with a genetic disease when considering reproductive decisions. Prospective parents may choose not to bear children or may take special steps to monitor their children as a consequence of information obtained about diseases that are more likely in their children than in the general population.

DISCLOSURE TO INSURANCE COMPANIES

The insurance industry is the second largest user of medical information in the United States, after the Federal Government (Baskin, 1978). Both life and health insurance companies use medical information in order to assess the probability of health events for those who are insured. There is a great deal of variation between individual firms in the amount of information required to accept an applicant.

Health Insurance Companies.—One hundred ninety million people in this country had some form of health insurance coverage in 1983 (Health Insurance Association of America, 1984), and many people consider health insurance to be a necessity. The majority of health insurance policies are group policies, received in conjunction with employment. These group health policies do not consider the health risks of the applicants to determine their insurability or their premiums. However, claims made on preexisting health conditions are exempted for a period usually **of 30** to **120 days** (Health Insurance Association of America, 1984). The access of insurance companies to genetic patient data, therefore, does not seem to be an **issue for most group health insurance coverage .**

Individual health insurance policies, however, are similar to life insurance policies, since they both use medical information to determine the premiums. Group health insurance policies, generally used in employee benefit packages, usually require applicants to sign a blanket waiver permitting access to their entire health record, including family history and any genetic patient data.

The people who purchase individual policies include those over the age of 65, the self-employed, and workers in small businesses. The unemployed do not qualify for group insurance and usually cannot afford individual policies. For the remainder of this paper, the term "insurance" will encompass both life and individual health insurance.

Life Insurance Companies.—Most life insurance companies require an applicant to answer several questions about his or her health on an application form, and then if the answers warrant, and if the coverage sought exceeds a certain amount, they may require the applicant to release his or her medical records, submit to a medical examination, or both. The results of these medical findings and other data are then used to determine the life insurance premiums for an individual, or whether the person is insurable at all. Some of the questions are related to conditions with a genetic component, such as sickle cell disease, and if an applicant reports or displays the symptoms it is unlikely that he or she will be insured. Likewise, an applicant may be asked about the presence of heart disease, high blood pressure, or stroke in his or her immediate family, and an affirmative answer would increase the risk factors involved, even though the genetic basis of these diseases is not clear. The use of this genetic patient data raises several ethical questions that are not new, but the potential increase in the amount of genetic patient data in the future may increase the significance of these issues.

Risk Classification. -Insurance companies generally use several factors to determine an individual's insurance premium, such as gender, occupation, weight, and blood pressure (Cummins, et al., 1983). Recently, some insurance companies have begun using lifestyle factors, such as one's smoking or exercise habits, in assessing insurance risk.

***Controllable Risk* Factors.—Smoking is considered largely a voluntary activity, controllable by the individual, with strong actuarial evidence of significantly reduced life spans. It is also generally accepted that the primary health effects from smoking (e.g., cardiovascular disease, emphysema, and lung, esophageal,**

and bladder cancers) can be caused by smoking. Perhaps the major drawback of using this type of lifestyle information is that it is self-reported and, therefore, not verifiable; since there is a price incentive to report that one is a non-smoker, an applicant may not be truthful.

Diet is also a known risk factor for development of certain types of diabetes, arthritis, and susceptibility to colonic and breast cancers. Alcohol ingestion is associated with liver cirrhosis, esophageal and stomach cancers, and more than a dozen neurological syndromes.

Uncontrollable Risk Factors.—Until a 1983 Supreme Court Decision, it was common practice in the insurance industry to use the gender of the applicant to determine the premium. While that practice continues in the underwriting of individual policies, it is no longer allowable in group health, or employee benefits, policies. (I. Katz Pinsler, ACLU, personal communication, 1984). In contrast to lifestyle factors, one's gender is genetically determined and is not under one's voluntary control, but there is strong actuarial evidence that women tend to live longer than men. Gender is verifiable, which makes it relatively easy to use as a determinant. Some actuaries, however, question the use of gender, claiming that other factors such as smoking, lifestyle, work habits, or competitive behavior maybe the cause of the mortality differences (Cummins, et al., 1983 p. 86), *(Business Insurance,* 1981).

The race of an applicant is not used to determine the premium, although the criteria are similar to the case of gender: one's race is not under one's own control, but although there is actuarial evidence for mortality differences between races, it is difficult in practice to identify distinct races because of the degree of racial mixing. Insurance companies argue that the actuarial differences between races are due to socioeconomic differences and not to race, per se, and that these factors are already considered in the actuarial process. Also, several States have prohibited the use of race in insurance underwriting (Cummins, et al., 1983 p. 90).

Some factors with genetic components are used to determine the insurance premium, such as a family history of heart disease. The criteria for using genetic patient data are similar to those for race and market since one's genetic complement is not voluntary. At present, however, most genetic diseases cannot be verified before they are expressed.

Efficiency and Equality.—Insurance companies, as profit-maximizing firms, have an incentive to use any readily available genetic patient data because it will allow them to function more efficiently in the free market. By using this information, they will be better able to identify high-risk applicants and thus be able to charge them proportionately higher premiums.

The adverse selection model, described below, provides one explanation for why insurance companies might wish to use genetic patient data in the underwriting process. In an insurance market, when there is no distinction made between the risks of the applicants, there is a tendency for those who know they are at risk to purchase the highest coverage they can afford. With more of these high-risk clients, insurance company costs will increase, because the company will be paying more claims. The increase in costs tend to drive up the insurance premiums, causing low-risk clients to leave, this results in a pool of high-risk clients, paying high premiums. If another type of insurance were available that differentiated applicants on the basis of risk, insurance companies could make a profit by offering it as an option (McGill, 1984). The use of genetic patient data could help insurance companies counter this adverse selection phenomenon which can lead to high rates.

The question of fairness remains, however, and the crux of the issue is whether it is more fair for those individuals with high risks to pay proportionately higher rates or for all individuals to pay the same rate, regardless of risk. In the first case, market forces will act to differentiate people on the basis of the risk they present to the insurance company, and may lead to groups of individuals unable to purchase insurance at an affordable price. This type of situation may seem fair when it concerns something over which an individual has some control, such as one's smoking habits, but the fairness issue becomes more difficult when it involves something over which one has no control, such as one's genetic complement.

In the latter case, where everyone pays the same rate, low-risk individuals would be subsidizing high-risk ones. In either case, one group will be harmed, and society needs to determine whether the low-risk or high- risk individuals will bear the burden. A compromise could be made using the U.S. Social Security system as a model. In this system, contributions are not actuarially equal to benefits, but the level of benefits is related to the amount contributed (Cummins, et al., 1983).

Impacts of Using Improved Genetic Patient Data.—Increased screening for genetic diseases could lead to numerous groups of individuals that are substandard risks, uninsurable, or who must pay prohibitively high rates. At present, diseases or health conditions that already exist carry more weight in the underwriting formulae than those conditions which are just statistical probabilities. If reliable genetic patient data were

available at low cost to use in insurance underwriting, however, more weight might be placed on them. For example, if an applicant has expressed polycystic kidney disease, he or she is likely to be denied insurance. However, since this disease is not expressed until later in life, an individual can carry the gene for the disease and still obtain insurance, since there is no way to detect the gene at present. If the gene could be detected at an early age and one could say with high certainty that a person would develop polycystic kidney disease, then such tests might be used to determine insurability. Further questions arise concerning the use of tests that are under development, are not perfectly accurate, or are prohibitively expensive. For example, if a test were developed which indicated the presence of a gene but not whether it would result in disease,[5] should the results of the test be used in the underwriting process? Three States, Florida, Maryland, North Carolina, (Case, Health Insurance Association of America, personal communication, 1984) already specifically prohibit health insurance companies from discriminating against sickle cell carriers.

The increased use of genetic patient data in the underwriting process has significant legal implications. Since several genetic diseases are linked closely with race, (see table B-1) if an insurance company uses genetic patient data to compute the health risks of applicants, it would have a disparate impact on the affected races. As genetic markers become more refined, it may become increasingly difficult to separate the prevalence of specific genetic diseases from race. Therein lies a potential conflict with current or future civil rights laws.

The role of Federal and State Governments in constraining access to genetic patient data may increase in proportion to the amount readily available. Patient protection will be afforded by case law, but some aspects of how genetic patient data are specifically handled (in contrast to other personal or medical information) may depend on new Federal or State regulations. Public policy on genetic patient data turns, in part, on whether it is classed as a basis, like race, for civil rights protections. As the availability of genetic patient data grows, pressures to use it and disclose it to third parties will also likely increase. Legislatures may wish to consider new laws to redress misapplications or to cover areas not clearly defined in case law.

DISCLOSURES TO EMPLOYERS

Because of the significant costs of occupational illness— including the time lost from work, the cost of training replacements, and increased health insurance rates—a profit-maximizing company has an incentive to reduce the incidence of work-related disease as long as the costs of the reduction are lower than the costs of the disease (Murray, 1983).

Because the expression of a genetic disease is frequently thought to be determined by a combination of genetic and environmental factors (Harsanyi, 1981), companies may have the ability to change specific environmental factors which otherwise enhance the possibility of disease expression. Availability of genetic data on employees could then lead to companies assisting those employees in remaining healthy.

But what if the cost of the disease is higher than that of the reduction? Or if there is no known way to reduce incidence? Or if a company is disinclined to institute changes due to either inconvenience or cost? The use of genetic patient data under these circumstances could lead companies to discriminatory hiring, promotion, or lay-off policies. In this light, the question once again arises as to whether companies should have general access to genetic patient data.

Title VII of the amended 1964 Civil Rights Act, and sections 503 and 504 of the 1973 Rehabilitation Act govern employment rights. The former prohibits employment discrimination on the basis of race, color, religion, sex, or national origin. The latter prohibits discrimination against otherwise qualified handicapped individuals by employers who are Government contractors or recipients of Federal assistance.

Currently, the term 'handicapped individual' is defined in section 503 as "any person who: 1) has a physical or mental impairment which substantially limits one or more of such person's major life activities, 2) has a record of such an impairment, or 3) is regarded as having such an impairment." Equally, in section 504, an employer receiving Federal financial assistance may not make preemployment inquiry about whether the applicant is handicapped or about the nature and severity of an existing handicap unless a preemployment medical examination is required of all applicants and the information obtained from the examination is relevant to the applicant's ability to perform job-related functions. Both sections serve to limit the use of discriminatory preemployment examinations and tests, but it must nevertheless be determined whether genetic trait is a handicap and whether screening procedures are job related.

These statutes indicate that individuals are not to be discriminated against on the basis of some immutable characteristics and that their abilities are to be judged on an individual basis. Since genetic screening could result in employment discrimination against groups of individuals with particular inherited traits, one question that arises is whether such discrimina-

[5] Since most diseases are due to a combination of genetic and environmental factors, genetic tests may eventually prove to be mostly of this type

Table B-1.—Genetic Diseases Found in Higher Prevalence Among Specific Racial or Ethnic Groups

Condition	Prevalence
Amyloid nephropathy associated with familial Mediterranean fever	1:3,000Sephardic Jews
Aspartylglycosaminuria	70-100 cases in Finland
Cystic fibrosis	1:2,000 Caucasians
Diabetes mellitus, type 2 (insulin-dependent, ketosis-resistant)	1:130 Caucasians, uncertain in blacks
Dubin-Johnson syndrome	1:1,300 Iranian Jews
Essential fructosuria	1:130,000; more common in Jews
Galactosylceramide lipidosis (globoid cell leukodystrophy; Krabbe's disease)	1:50,000 in Sweden
Gaucher's disease, type I	1:2,000 U.S. Jews
Glucose-6-phosphate dehydrogenase (G6PD) deficiency; multiple allelic disorders, including mild A-type and severe Mediterranean type	A-type: 1:11 U.S. blacks (males) Mediterranean type: common in Africa, Middle East and other Mediterranean countries
Gyrate atropy of the choroid and retina	1:50,000 in Finland
Hereditary fructose intolerance	1:20,000 in Switzerland
Hereditary spherocytosis, several types	1:5,000 Caucasians
Hermansky-Pudlak syndrome	1:60,000 Caucasians 1:5,000 Puerto Ricans
Intestinal lactase deficiency	1:10 Caucasians, majority of Asians, Africans, and U.S. blacks are affected
Niemann-Pick disease	1:25,000 U.S. Jews
Nonketotic hyperglycinemia	1:250,000 in United States 1:12,000 in Northern Finland
Occulocutaneous albinism, tyrosinase-negative type	1:39,000 Caucasians 1:28,000 blacks
Occulocutaneous albinism, tyrosinase-positive type	1:37,000 Caucasians 1:15,000 blacks 1:150 in certain American Indians
Pentosuria	1:2,500 Eastern European Jews
Primary gout: idiopathic	1:500 in Western populations 1:50 in American males by age 50 1:10 in males in some Polynesian groups 1:25 in females in some Polynesian groups
Sickle cell anemia	1:500 U.S. blacks (newborns)
Tay-Sachs disease	1:3,000 U.S. Jews
Thalassemia, multiple allelic disorders	High frequency in Mediterranean, African, and Asian populations
Tyrosinemia, type I (hepatorenal tyrosinemia; tyrosinosis)	1:10,000 French Canadian isolate
Variegate porphyria	Common in South Africa; rare in other parts of the world
Xeroderma pigmentosum, multiple types involving multiple gene loci	1:25,000 in Egypt

SOURCE: Stanbury, 1983; as amended by Bowman, personal communication, 1984.

tion is prohibited by these two acts (OTA, 1983). If they are judged not to prohibit genetically based discrimination, another question raised is whether additional federal legislation will be forthcoming.

The 1970 Occupational Safety and Health Act (OSHA), which requires employers to maintain a workplace free from recognized hazards, does not specify the means by which that requirement can be met. For example, it neither supports the argument that genetic testing is required nor that genetic testing is prohibited. Although the results of genetic testing could have an adverse affect on particular employees, it certainly cannot be classified as a "hazard" (OTA, 1983). Yet genetic testing might become the basis for employment discrimination, or harm to employees.

In this light, it is significant to note that it is common practice for employees to sign a blanket waiver allowing the company to gain access to all medical records it deems necessary. Employees generally "have little genuine expectation of true confidentiality as to employment medical records" (OTA, 1983). Any duty to the confidentiality of the patient is based on a physician-patient relationship, and the traditional view is that a physician-patient relationship does not exist be-

tween an employee and an employer-provided physician. Some courts take a view that the existence of a physician-patient relationship is dependent on the context of the health care provided. If the physician-patient relationship does not exist, neither does the duty of confidentiality, and so the company generally may have access to the medical records of its employees.

There are also few common law restrictions on the disclosure of genetic patient data to parties outside of the company, except for several State and Federal restrictions. For example, California requires employers to establish procedures to protect the privacy of medical records, and records may not be released without the consent of the employee. Because of the potential harm to the employee arising from disclosure, legislators may wish to anticipate the outcome of the increased use of genetic information by employers.

Unauthorized Access.—Because of the use of computers to maintain health records, there has been a growing concern for the security of the information, especially in light of the reports of computer crime. These concerns are not unique to the health care field, since every major sector of the economy is relying more on the computer for the maintenance of records. Genetic patient data may not be as obvious a candidate for computer theft as would be valuable trade secrets, but patient records at Memorial Sloan-Kettering Cancer Center have already been broken into (Marbach, 1983), and so the possibility of unauthorized or inadvertent access should not be discounted as greater amounts of genetic patient data become stored.

One solution to the problem of unauthorized access is to remove any identifying data from the record and keep it in a separate file. Then codes could be used to match the individuals to their records. Another solution is to extend the concentric circle model of privacy to include the genetic patient data stored in computer files. The information could carry different access codes, so that information could be accessed only by those physicians who need to know. Different individuals would therefore have access to different levels of private information, but this would not obviate the need for patient control of disclosure of information to third parties (Walters, 1983). These safeguards, while protecting the privacy of individuals, might also have the detrimental effect of making it more difficult for physicians to use the information in the medical record. The experience of research on Huntington Disease suggests that it is possible, by careful attention to data entry and access restriction, to provide aggregate data while protecting individual privacy (Wexler, personal communication, 1984).

DISCLOSURE TO THE GOVERNMENT

The government will likely play an important role in issues relating to genetic patient data both as a significant user of information and as a body acting to control the access to that information.

The major objective of government in using medical information is the protection of the public health. For example, by collecting statistics on the frequency and incidence of various diseases, the government perhaps can take measures against those diseases in the future, perhaps by mobilizing health care efforts in particular areas. Other likely government uses of genetic patient data include:

- providing information about medical costs,
- developing policies to better allocate health resources, and
- identifying diseases which merit additional research.

For these purposes, the identity of the individual is not important, and so all identifying pieces of information can be culled from the record. For other purposes, such as tracking individuals with specific genetic diseases or doing epidemiological research, however, it is important to know the identity of those at risk. In the interests of privacy and security, the records may be coded and the identifying information may be stored in a separate file, but the identity of individuals must still be accessible. In this instance, privacy can be retained by authorizing only one, or a few, disease centers to follow individual patients.

Because of the growing amount of information collected and used by the Federal Government, and because of improvements in information storage and retrieval technologies in the foreseeable future, Congress passed the Privacy Act of 1974 to set a policy for the appropriate use of personal information. The Act states that "The right to privacy is a personal and fundamental right protected by the Constitution of the United States, " and that in order to "protect the privacy of individuals identified in information systems maintained by Federal agencies, it is necessary and proper for the Congress to regulate the collection, maintenance, use, and dissemination of information by such agencies" (Privacy Protection Study Committee, 1977). The Act describes in detail the conditions of disclosure and access, as well as agency requirements and rules. The Act forbids the disclosure of any records to any person or agency, except with a written request by, or with the prior request of, the individual to whom the record pertains. Violations of the Act can lead to a civil liability.

The Federal Government is involved in providing funding for genetic testing and counseling, thus assisting in the process of collecting genetic patient data. The government can also serve as an effective forum to discuss the ethical, legal, economic, and social aspects of genetic information. Since there are many different groups involved in balancing the issues of privacy and access, the Federal Government can ensure that these issues are included in the decisionmaking process.

The States also have the authority to compile and store genetic patient data that is of potential benefit to the public health (Reilly, 1977 pp. 250-252). Several States have written laws that regulate the type of information that can be collected and the procedures through which disclosure can be made (Reilly, 1977 pp. .252 -256) The States also have control over the business practices of various industries including insurance companies, and they may determine the propriety of using genetic patient data in different employment and underwriting situations. Some States have already forbidden the use of such data as gender, age, handicaps, or other impairments in the underwriting process (Cummins, et al., 1973).

General education is an issue of Federal, State and local interest, necessary so that all people can have an understanding of genetics sufficient to understand the complex issues of genetic patient data (President's Commission, 1983; Rowley, 1984). Some schools have responded to this need by making genetics a major focus of their biology courses. Genetic education is an issue for health care providers, as well. The teaching of genetics occurs primarily' during the first 2 years of medical school, with little integration of genetics into the practical side of clinical training (Rowley, 1984). As the technology for identifying genetic disease improves, it is important that physicians become aware of that technology and how to use it with patients.

Conclusion

Public policy on genetic patient data is centered on determining the rights of privacy and access, pitting individual autonomy against relatives' or third parties' needs for information. The legitimacy of others' needs are determined by the potential benefits to relatives, health providers, insurers, employers, or the general public compared to potential harm to the patient from disclosure, Several factors are included in such assessments, including the seriousness of the genetic condition, the genetic relationship between interested parties, and the probability of preventing harm or promoting good by disclosure. When no genetic relationship exists, as in the case of insurers and employers, issues of fairness arise. Continuing public scrutiny may be instrumental in the evolution of deciding on a hierarchy of conditions and people for whom disclosure of genetic patient data is important (Rosenfeld, 1984).

Public policy on genetic patient data attempts to control access so that individual privacy is protected. This effort may include support of data storage methods that are coded so that epidemiologic research and research priority assessment may be performed without jeopardizing individual privacy. Legislation may be required to guide genetic data collection agencies in what constitutes appropriate disclosure of information and to act as a deterrent to unauthorized access. Public policies may be required that would strengthen individual's control over access to their genetic data. In contemplating new legislation, care must be taken to ensure that controls are not so strict that the genetic patient data cannot be used for legitimate and lifesaving purposes.