Chapter 8

# The Worker as a Person: Individual Uses of Genetic Information

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# Chapter 8

# The Worker as a Person: Individual Uses of Genetic Information

Our genetic identity is more than DNA sequences. Our genes carry much that is relevant to our past and our future. They also carry secrets. Everyone possesses a minimal number of deleterious genes, that may or may not be expressed, depending on their location, their phenotypic expression, the environment to which they are exposed, and the life choices of those who bear them.

Most often, individuals learn about their genetic identity in the context of family planning. Prospective parents may choose to undergo diagnostic tests so they can increase their reproductive choices. In other cases, individuals learn about their genetic identity because they, or a relative, are diagnosed with a genetic disease or syndrome. But in both cases, the individual requests to participate in testing and subsequent counseling, and understands, even if in rudimentary terms, why the tests are being done. At the least, the fact that tests are conducted in a medical setting provides a context in which certain assumptions and expectations can reasonably be held by the person being tested. These factors might be different if the workplace becomes the background for receiving genetic information.

New technical capabilities to diagnose and predict genetically based disease have opened new pathways for informed decisionmaking about ourselves and our family's health. But they have also created moral, ethical, and psychological dilemmas for which there are no easy solutions. In addition, genetic monitoring and screening tests often convey a probability, but not a certainty, that disease will appear, introducing difficult uncertainties into the lives of those tested. Other chapters in this report address the issues surrounding the application and use of tests (both monitoring and screening) in the employment setting. This chapter will address issues faced by the individual who undergoes testing-not as a worker—but as a person and a family member. It discusses the role of genetic information in family life and personal health and the need for sufficient and appropriate counseling for those who find themselves, or their families, at risk.

## MONITORING v. SCREENING: ISSUES FOR THE INDIVIDUAL

Three approaches have been proposed to consider the various issues in genetic monitoring and screening (19). The frost is fatalistic, where the existence of a particular genetic vulnerability is recognized as a quirk of fate which could affect anyone and for which society cannot be held responsible. This approach most closely resembles the state of public thinking until recently. As a society, we are quickly moving away from this perspective.

The second approach is individualistic; i.e., society assists the individual to better understand the problem and find the best means of dealing with it. The burden, however, is on the individual to act or be acted on. Screening an individual for genetic traits and diseases, and removing the individual from an allegedly hazardous environment on the basis of test results reflects the individualistic approach. But, as discussed in chapter 5, there are technical and practical constraints to this way of thought because of the limits of the tests themselves and the uncertainty of cause and effect. Despite the technical constraints of testing, the individualistic approach is currently taken in the clinical genetics setting, the routine environment for genetic tests.

The last approach is social welfare activism. It applies the societal principles of justice and equity to the genetically afflicted individuals. This view holds that where conditions are found to be unequal, or natural differences contribute to inequalities, they should be rectified to benefit the least well-off person. The use of genetic monitoring to identify areas of potential risk for all individuals reflects the concept of social welfare activism. In this scenario, actions taken on the basis of test results are taken on the group, not the individual. No one is singled out.

The difference between the individualistic and social activism approaches lies in the locus of burden and the implications for action. In the individualistic approach, which involves screening, the burden of dealing with the test results is placed on the individual as a worker and a patient. In the social welfare approach, which also includes moni-

toring, the burden rests on the company to take action by lowering or removing risks. Testing becomes a source of surveillance data.

Whatever avenues society takes in applying genetic monitoring and screening tests to workplace decisions, it can never be overlooked that the focus of the tests—the person—is being provided with information which may have a significant impact on decisions unrelated to employment; these include marriage, procreation, and lifestyle. While positive test results could be the end of the story for the employer (having decided not to hire, to relocate, or to fire the individual), they are likely to be the beginning of the scenario for the individual, who must now decide what the findings mean in his or her private life.

## THE NEED FOR SUFFICIENT AND APPROPRIATE COUNSELING

For many individuals, even considering whether to undergo genetic monitoring or screening constitutes a life crisis because of the possible outcomes. If the results are positive (for the trait, disease, or genetic change), the crisis obviously is exacerbated. How the results will affect the individual has much to do with the individual's own frame of reference, but also with the implications of the condition and its prognosis.

Psychological issues permeate every aspect of genetic consultation. Information received can be ego-threatening or even life-threatening, as individuals find that they are "flawed, "imperfect, " "defective," "inadequate,' or "abnormal, ' and may have the potential of transmitting these "flaws' to their progeny (16,17). How the information is obtained, communicated, retained, and eventually used by the person being tested involves a "series of complex, multidimensional processes with major rational and nonrational components" (17). In addition to the intrapsychic consequences of receiving genetic information, there are potential impacts on family. Genetic information affects not only the individual, but also the spouse, parents, grandparents, siblings, and children. Social and psychological stress, as well as future financial and emotional burdens, can strain family functioning (29).

In addition, genetic conditions found through screening are permanent and chronic and may evoke

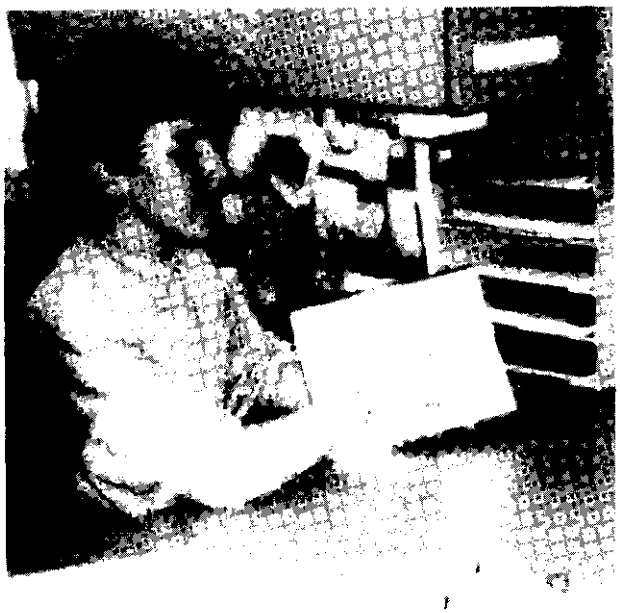

*Photo* credit: Diane Baker

**A genetic counselor showing a chromosome chart to a client. Genetic counseling may assist individuals and families cope with positive test results.**

labeling. The continuous, ever present diagnosis of genetic disease or potential for disease may elicit "chronic sorrow" (24) in those affected. In addition to coping with their own uncertain future, individuals may experience guilt or grief if they find they have unknowingly passed a deleterious trait to their offspring.

Obviously, the psychological impact of a positive diagnosis varies with its severity and treatability, and the fact that different families will react uniquely to similar situations. Support, counseling, and followup are likely to assist individuals and their families in coping with positive test results. The knowledge and skills of a properly trained counselor can help the individual understand the diagnosis, recurrence risk, prognosis, relevant preventive and therapeutic measures, and also aid in communicating important information to other family members. When these goals are accomplished, genetic counseling is usually perceived as a valuable experience by the counselee and the counselor (22).

When it is not possible to make a specific diagnosis, or to give an accurate recurrence risk or more than a very general prognosis, as will be the case in many predictive tests, interactions between the testee and the tester are even more complex. Until research progresses, it is likely that non-

specificity of diagnosis and prognosis will predominate in workplace genetic monitoring. Employers undertaking such programs should anticipate the complexity of interpretation and communication of test results.

## *Pretest Counseling*

Professionals in the fields of health and education are usually the first to see families who are seeking information about genetics. Helping a client to cope with the question "why is this happening to me?" is one of the objectives of pretest counseling. Another objective is helping a client understand the test—what it is and is not, as well as why it is being done. When employees are tested at the workplace by someone insensitive to counseling objectives, the workers may be confused throughout the entire process.

In routine genetic counseling, the client discusses why he or she chooses to be tested and discusses with the counselor the implications of the possible outcomes. The counselor prepares the individual for both positive and negative test results. It is also the time to discuss risk reduction strategies, if relevant.

### Understanding Risk

One of the genetic counselor's tasks is to communicate risk to the client-a job not easily performed (13). A decision to be tested will be influenced by a person's perception of the chance that the test will be positive. The interpretation of numerical risk varies depending on: prior perception of the magnitude of the risk; anxiety state of the client at the time of the test; familiarity with the outcome (whether there is an affected relative); how treatable the condition is; and belief that the outcome with which the individual is familiar is representative of all such outcomes (15).

The perception of risk may be a more important determinant of decisionmaking than the actual risk. The way risks are posed by the counselor may, in fact, influence the client's choices. When confronted with the risk of genetic disease in their offspring and when making reproductive decisions, people tend to place greater weight on their ability to cope with a disabled or fatally ill child than on precise numerical risks. For example, for some couples, a risk of 10 percent could be perceived no differently than a risk of 50 percent if they believe that they cannot cope with the situation. In prenatal counseling, regardless of actual risk, parents overwhelmingly perceive the chance of recurrence as either O or 100 percent-it either will or will not happen. By processing rates in this way, individuals simplify probabilistic information and shift their focus to the implications of being at risk, and the potential impact of what could occur ***(20).***

***In*** addition to the subjective factors that influence the interpretation of risk already discussed, the understanding of risk in arithmetic terms is usually deficient. Comprehension of the concepts of probability and risk will influence the client's understanding of the genetic information provided by testing (16). In a Maryland study of 190 predominantly White, middle-class women, over one-fifth thought that "1 out of 1,000" meant 10 percent, and 6 percent thought it meant greater than 10 percent (5).

The way risks are framed also influences choices (21). The decision to have a genetic screening test can be different if the risk is presented as a 25 percent chance of having an affected child rather than a 75 percent chance of having a normal child. Presenting risks in personal terms may improve the chance that action will be taken (13).

Most studies of counseling have focused on cases where the patient already has an affected child or relative and is familiar with the disorder. Little is known about the effect of counseling prior to genetic screening in people with no previous family history of the condition for which they are being tested. It is likely that their misperceptions could be great. Pretest counseling is all the more imperative in these cases, as is the need for informed consent.

### Obtaining Informed Consent

The following text presents the routine consent process in contrast to that which we will find in the workplace. (See ch. 6 for further discussion of informed consent.) In the routine clinical genetics setting, very few situations arise in which genetic monitoring or screening can be performed by a health care provider without informed consent. Before any invasive procedure (including the taking of samples such as blood, urine, or saliva) the individual should be informed of the following:

- purpose of the test,
- risk of the test itself,
- validity of the test (the possibilities of false results),

- implications of a positive result (medical and social),
- nature of the condition for which the test is being conducted,
- options available to reduce the burden of disease in the event of a confined positive test result, and
- alternatives if the individual decides not to have the test.

Most of these, or analogous elements, are specified in the informed consent statutes of 21 States (l). Informed consent is not obtained when a disclosure is incomplete, constructed to prejudice the subject toward a particular action, or obtained under pressure.

The process of obtaining informed consent in the medical sense may not be practiced in the workplace. Consent may be obtained for using the results to make hiring and employment decisions but the future or current employee may not receive the information needed to obtain explicit consent for medical intervention.

### *Posttest Counseling*

When attending a genetics clinic for reasons other than prenatal genetic screening, people have historically come because they have had an affected relative, usually a child. They tend to be familiar with the disorder. The affected relative, rather than a test, served as the indicator of potential disease for others. As an increasing number of genetic screening tests are administered to healthy individuals with no apparent family history of genetic disease, counselors will have to spend more time describing the disorder to those with positive test results.

Studies have shown that test results should be reported in person, by the same person who provided the pretest counseling (9). If the test results are positive, prior contact may have alerted the counselor as to whom else should be informed, whose help might be needed on behalf of the client (i.e., financial or emotional support), and important information about the client's lifestyle and family (as well as financial and insurance information).

Followup counseling and support is also strongly advised. News of a positive result impedes a person's ability to accept advice on both emotional and practical levels. Faced with positive results, most individuals are unable to take advice until they overcome the shock and possible denial that their fate or their children's fate could suddenly shift in a negative direction. Information about treatments and the importance of changing lifestyle is best assimilated several days after the test results are communicated. Focusing on medical facts at this stage could convey to the individual that the psychological issues he or she is dealing with are unimportant or irrelevant (38).

Even in the best of all worlds, where consistent counseling has been provided all through the process, the effectiveness of counseling is sometimes questionable. An analysis of nine studies on counseling published since 1970, concluded, "many parents of children with a genetic disorder have an inadequate understanding of the genetic implications of the disease, even after one or more genetic counseling sessions" (10). One survey found that more than half of the 87 percent of people who came to a genetic counseling center with inaccurate knowledge of risk were still misinformed after counseling (13).

The task of communicating genetic information is formidable. Counseling programs are continually trying to educate counselors to improve the process (35). A major impediment to satisfactory counseling has been a profound lack of understanding of basic genetics. Anyone administering tests necessarily takes on the role of educator as well as practitioner and examiner.

## THE ROLE OF GENETIC INFORMATION IN FAMILY MANAGEMENT AND PERSONAL HEALTH

A person's genetic constitution (genotype) determines the broad limits of his or her potential, whereas the expression of that potential (phenotype) is dependent in an important way on the environment with which the genes interact. The assumption that there is always a one-gene-disease relationship is fallacious. There are numerous variables such as general health, diet, medication, and stress that contribute to or interact with the genetic trait, in addition to workplace exposures, to produce a disease state.

The harmful manifestations of some genetic diseases can be prevented or ameliorated by the administration of drugs or special diets, or by the

elimination of harmful environmental agents. To be optimally effective, intervention must take place early, frequently before symptoms of the disease appear. Predictive tests have been unavailable for most single-gene diseases, but with the use of recombinant DNA technology many are being developed (see ch. 5).

For most genetic diseases, the basic defect is not known and effective interventions are not yet feasible. Although linkage studies or direct DNA analysis will eventually reveal the defect, there will be long delays between the time the gene is located and the time when effective interventions are available. In the meantime, predictive tests for those at risk could be developed and widely disseminated. Healthy individuals could learn of their fate as potential patients and face several options, depending on the prognosis for the disease and the availability of effective intervention.

### *When Intervention Is Available*

A considerable amount is known about the pathogenesis of some multifactorial conditions, such as diabetes mellitus, coronary artery disease, and lung cancer. If it were possible to identify individuals with genetic susceptibilities to these conditions, the pathogenic process might be interrupted (if the person at risk adopts risk-reducing behaviors). In contrast to highly penetrant single-gene disorders, however, it is doubtful that all persons found to have a susceptibility-conferring genotype would eventually manifest the disease, even if they possessed other predisposing alleles or were exposed to harmful environmental agents. Unless one can be certain that disease will appear, potentially harmful interventions should not be used on those who may never become sick in the first place. Avoidance of dispensable habits, however, such as smoking or a high fat diet, would be safe plans of action (see box 8-A).

Individuals found to be at risk for non-insulin dependent diabetes could be counseled about the importance of weight control. Counseling people found to be at risk for colon cancer to increase fiber intake, or to have periodic colonoscopic testing could ensure early treatment. Those at risk for melanoma could be advised to protect themselves from sunlight.

In all these cases, individuals can be informed of the likelihood that specific actions they can take, could modify the prognosis delivered with the test results. When positive test results are based on monitoring, rather than screening, the individual's choices are not as clear (see ch. 4). At that point, the patient as a person may wish to be removed from the potentially hazardous exposure, but the patient as a worker may have no choices.

### *When Intervention Is Unavailable*

For many disorders, neither drugs nor diets nor lifestyle changes have yet been found that markedly improve the outcome. This greatly complicates the personal burdens of threatening medical information. Examples of such disorders with a late onset are Huntington's disease and Alzheimer's disease (see box 8-B). Other disorders can be treated with some benefit, but the outcome is not always good and the management of the disease may be costly. Maple syrup urine disease, hemophilia, bipolar affective disorder, and schizophrenia fall into this category.

The psychological sequelae of facing the uncertainties of untreatable illness are devastating. There is a growing body of literature related to coping behaviors associated with testing positive for HIV antibodies. One of the most psychologically unacceptable notions which confronts the individual at risk is to be the passive victim of a totally random event (36). Another aspect of detecting a late onset disorder is the possibility of self and social stigmatization, and the increased opportunity for discrimination (16).

### *When the Test Results Are Inconclusive*

***In the*** case of screening for genetic disease, most tests are fallible. Some of the problems are specific to the method employed and some to the laboratory performing the test. Others result from genetic heterogeneity and incomplete penetrance. Failure to correctly interpret monitoring and screening test results poses a significant problem for the patient.

In the case of genetic monitoring, the uncertainty is probably even more pronounced. Because of the lack of causal linkages between exposures and clinical prognosis, the clinician is left with little on which to base a prognosis. If the results reveal chromosomal damage, there is little reliable and valid information available that would allow individuals to make informed choices. For example, they can be told that there appear to be causal linkages between cancer and their condition, but that there is

### *Box 8-A—Screening for Coronary Artery Disease*

Coronary artery disease (CAD) is a major public health problem. Myocardial infarction, secondary to CAD, causes 30 to 35 percent of all deaths in men between ages 35 and 50 and is responsible for more than half a million deaths per year in the United States. CAD results from atherosclerosis, a slow, progressive disease of the arteries that begins early in life and may go undetected until the first heart attack which maybe fatal.

A strong association between hyperlipidemia and the risk to develop CAD has been demonstrated. There is evidence for the existence of three monogenic forms, as well as of polygenic and nongenetic forms, of hyperlipidemia. Familial hypercholesterolemia, familial hypertriglyceridemia and familial combined hyperlipidemia are transmitted as autosomal dominant traits and are well-established entities. In most cases of autosomal dominant transmission of CAD, the individual has symptoms that lead to the diagnosis.

Several different genetic factors have been associated with CAD. Only 1 percent of those classified as hyperlipidemic have a clear-cut monogenic cause. Nongenetic factors, e.g., cigarette smoking, high cholesterol diets, obesity, physical inactivity, stress, and diabetes mellitus, may also contribute to the disease state. Most cases, therefore, are heterogeneous or multifactorial and would be prime candidates for some type of predictive tests. Intervention could be started well before the appearance of heart disease.

Research using restriction fragment length polymorphisms has demonstrated an association between a 3.3 kilobase band and inherited apolipoprotein abnormalities that could predispose an individual to CAD. Tests at the DNA level may ultimately prove better predictors of CAD than lipid or apofipoprotein measurements. These tests may provide risk information prior to elevated lipid levels. When such tests become widely available, persons at risk could begin a prevention program including lowering dietary levels of cholesterol or taking drugs that bind cholesterol-like compounds in the intestine or inhibit cholesterol biosynthesis.

SOURCES: R.T. Acton, R. Bamberg, R.C.P. Go, et al., ''Utilization of Genetic and Other Laboratory Test Results To Predict and Reduce the Risk of Disease," *Proceedings of the Society of Prospective Medicine, 1988;* J.L. Goldstein, W.R Hazzard, H.G. Schrott, et al., "Hyperlipidemia in Coronary Heart Disease," *Journal of Clinical Investigation 52:1544, 1973;* A.G. Motulsky and H. Boman, ''Screening for the Hyperlipidemias,' Genetic Counseling, H.A. Lubs and F. de la Cruz (eds.) (New York, NY: Raven Press, 1977); J.M. Ordovas, E.J. Schaefer, D. Salem, et al., ''Apolipoprotein A-I Gene Polymorphisms Associated With Premature Coronary Artery Disease and Familial Hypoalphalipoproteinemia' ' New *England Journal of Medicine* 314:671-677, 1986; J. Stamler, "Epidemiology of Coronary Heart Disease," Medical *Clinics of* North *America* 57:5, 1973; G.L. Vega and S.M. Grundy, "Treatment of Primary Moderate Hypercholesterolemia With Lovastatin (Mevinolin) and Colestipol," ***Journal of the American** Medical Association 257:33-38, 19S7.*

no certainty that they are going to develop cancer. For individuals trying to cope with uncertain medical prognoses "(e.g., cancer, multiple sclerosis, or Hodgkin's disease) the loss of certainty in one's future (having a family, children, grandchildren, retirement, etc.) is often just as destructive to mental well-being as the certainty of death from a definitive prognosis.

## *When the Results Affect Reproductive Futures*

In the past 15 years, genetic screening tests have most frequently been used for prenatal diagnosis and family planning. Tests intended to detect disorders in offspring appear to be viewed differently than tests undertaken to identify personal risk. A 1986 Office of Technology Assessment (OTA) poll found that a majority of Americans who think human gene manipulation is morally wrong in the abstract approve of its use to save lives and heal sick children. In addition, a large majority of those polled (77 percent) say they approve of human genetic alteration to stop children from inheriting nonfatal birth defects or to reduce the risk of developing a fatal disease later in life (32).

Those identified as being carriers of autosomal recessive disorders (e.g., Tay-Sachs disease, thalassemia, and sickle cell anemia) currently have several options in family planning. They can proceed with an unmonitored pregnancy knowing that they have a 25 percent chance of having an affected child; if a prenatal diagnostic test is available, they can avail themselves of amniocentesis or chorionic villus sampling to determine whether the fetus is affected, at which point they can decide whether to continue or terminate the pregnancy; they can choose to become pregnant by alternative methods, or not to become pregnant at all.

Adequate and timely information is a key factor in helping families make their choices. Prospective parents need to understand the prognosis for an affected child before making a decision. For exam-

### *Box 8-B—Huntington's Disease*

Huntington's disease is a chronic, progressive, degenerative disorder, beginning usually between the ages of 30 and 50 years. It is characterized by uncontrollable, spasmodic movements in the face and extremities, as well as gradual loss of mental faculties, ending in dementia. The disease is lethal and incurable; death usually occurs on average 15 to 17 years after disease onset. The disease is transmitted as an autosomal dominant trait; offspring of an affected individual have a 50 percent chance of developing the disease. The test for the Huntington's gene is most often performed on an asymptomatic individual. If someone has the gene, that person will definitely develop the disease. Symptoms for the disease usually begin past the typical childbearing years, between ages 35 to 45.

The test provokes considerable anxiety among those at risk who elect to take it. Not all of those at risk elect to be tested, even though there is a 50 percent chance that they will receive good news. Prior to the availability of a predictive test for Huntington's disease, surveys indicated that between 56 and 85 percent of those at risk would avail themselves of the test. In a survey conducted after the test became available, less than 14 percent of the sample population at risk elected to take the test.

In another survey, 66 percent of the sample population at risk said they wanted the test. Of that group, 15 percent said they might commit suicide if the test were positive. Of the group that chose not to be tested, 30 percent feared they might be suicidal and therefore did not want their fears confirmed. For some people, uncertainty appears to be preferable to certainty.

A recent study on the psychological reaction of people being tested for the disease found no clear increase in psychiatric illness among people who tested positive for the Huntington's gene. People's reactions to their test results ranged from ''extreme joy and relief to disappointment, sadness and demoralization. " This study suggests that people cope well with this type of information if they are carefully screened, counseled, and provided followup care. In addition, those who test positive should be given appropriate long-term monitoring.

SOURCES: Office of Technology Assessment, 1990; based on C. Mastromauro, R.H. Myers, and B. Berkman, "Attitudes Toward Presymptomatic Testing in Huntington's Disease," *American Journal of Medical Genetics 26:271-282, 1987;* K.A. Quaid, J. Brandt, and S.E. Folstein, "The Decision To Be Tested for Huntington's Disease, " *Journa/ of the American Medical Association 257:3362 (letter), 1987;* B. Teltscher and S. Polgar, "Objective Knowledge About Huntington's Disease and Attitudes Toward Predictive Tests of Persons at Risk," *Journal of Medica1 Genetics 18:31-39,* 1981; A. Tyler and P.S. Harper, ''Attitudes of Subjects at Risk and Their Relatives Toward Genetic Counseling in Huntington's Chorea" *Journal of Medical Genetics 20179-188,* 1983.

pie, in one study, at least **89 percent of 333 couples identified as at risk for having children with Tay-Sachs disease used prenatal diagnosis (14). Tay-Sachs is a progressive, fatal disorder that results in death usually before a child's fifth birthday.** On the other hand, couples at risk for sickle cell anemia might not seek prenatal diagnosis, possibly because the disease is partly manageable and, therefore, many women would not abort an affected fetus (see chs. 3 and 5).

In the case of a late onset autosomal dominant disorder (e.g., Huntington's disease, or adult polycystic kidney disease), adults at risk face a double dilemma. Before the availability of predictive tests, individuals at risk (who knew of their risk status) could forego childbearing as the only way of avoiding passing on the trait. Now that those at risk can find out whether they will most likely develop the disease, they are presented with new options. If not at risk, they can freely reproduce without the burden of passing the gene to their children. If found

*Photo credit: Woody Guthrie Publications*

**Woody Guthrie: A famous American folksinger who died of Huntington's disease.**

*Photo credit: Diane Baker*

**An employee's genetic information can play an important role in his or her family planning.**

to be carriers of the gene, they can elect to have prenatal diagnosis to determine whether their offspring will also inherit the fatal gene. However, this is complicated by the fact that many do not know they are at risk until they have already had children.

The availability of these genetic screening tests is recent enough that very little is known about how high risk people who are tested deal with the psychological aftermath. Clearly, counseling and other support services should be offered in conjunction with any test.

### *When the Results Affect Other Family Members*

In the case of genetic monitoring, it is unlikely that positive results will directly affect other members of the existing family (with the exception of the unborn). Obviously, other family members can be secondarily affected by any consequences of potential or real deteriorating health of a loved one.

In genetic screening, there is a real possibility that test results will affect other family members. In the usual genetic counseling setting, the person being tested (the proband) is routinely advised of risks to other family members. For example, if the client is found to be a carrier for an autosomal recessive disorder, e.g., Tay-Sachs or sickle cell disease, the counselor informs the client that siblings each have a 50 percent chance of also being carriers. In most cases, the counselor suggests that the proband contact his or her siblings and suggest that they consult with their personal physician or come to the same clinic. The counselor cannot confirm that the proband has informed relevant family members. Unauthorized disclosure of medical information could result in legal action.

The issue of disclosure of medical information to others, e.g., insurers and employers, is discussed in chapter 6. Disclosure of medical information to relatives raises different issues. Not all families are emotionally and psychologically secure. Sibling relationships could impede full disclosure. Sharing highly personal medical information that involves reproductive and health futures may cause personal embarrassment or emotional stress for family members.

The question of duty to warn the proband's spouse also arises as a consequence of genetic screening. For example, a woman informed that she is a carrier of an X-linked condition might not wish to inform her husband or prospective husband that their male offspring will have a 50 percent chance of being at risk.

The President's Commission for the Study of Ethical Problems in Medicine and Biomedical and Behavioral Research took the position that physicians may release genetic information to relatives without the patient's or client's consent provided certain conditions are met. They are:

- reasonable efforts to elicit voluntary consent to disclosure have failed;
- high probability exists both that harm will occur if the information is withheld and that the disclosed information will actually be used to **avert** harm;
- identifiable individuals will suffer serious harm; and
- appropriate precautions are taken to ensure that only the genetic information needed for diagnosis and/or treatment of the disease in question is disclosed (26).

A different view was taken by the Committee for the Study of Inborn Errors of Metabolism of the National Academy of Sciences. It held that "under current law, genetic screeners would be ill advised to contact relatives without the screenee's explicit consent" (8).

## AVAILABILITY OF AND ACCESS TO GENETIC SERVICES

Tests are already available for a multitude of conditions with a genetic component. There are numerous tests available to diagnose a preexisting genetic condition in an individual or in utero. Several States require genetic screening for certain genetic conditions in the newborn period (e.g., phenylketonuria, sickle cell anemia) (30). In addition, tests are available to identify carriers of autosomal recessive conditions such as Tay-Sachs disease. Traditionally, these tests have been used almost exclusively within the disciplines of pediatrics and obstetrics.

In the next decade, it is estimated that tests will be available to identify genetic predispositions to certain disease, such as cancer or heart disease (31). In a 1987 survey of firms developing tests, half of the respondents felt that within 5 years, demand for genetic testing would outstrip current laboratory capabilities (31).

Comprehensive diagnostic, treatment, and management services are offered to high risk or symptomatic individuals and their families at approximately 20 clinical genetic service centers throughout the United States (33). Most (63 percent) are located at university-affiliated medical centers, with some centers operating satellite clinics in rural areas. However, genetic counseling services are not readily available to everyone, particularly those unable to pay for the tests themselves.

In examining the ability of workplace monitoring and screening programs to provide adequate followup services, the following should be considered. Genetics consultations tend to require longer office-visit time than most other specialties because of the need for detailed family histories and thorough physical examinations. Considerable time may be spent explaining the diagnosis for several family members as well as providing counseling (2). In addition, there are a number of other potential barriers-geographic, financial, linguistic, cultural, and educational-to the provision of followup services. Perhaps the greatest barriers to be overcome are those related to language and cultural differences (23). Bilingual genetic counselors will be needed in increasing numbers as more immigrants come to the United States.

*Photo credit: University of South Carolina School of Medicine*

**Genetics consultations can require longer office-visit time because of the need for gathering detailed information about the client. This genetic counselor is discussing the client's chromosome profile with her.**

As tests become available to a growing number of presumably healthy Americans, the administration of diagnostic tests and subsequent treatment for an increasing number of individuals will have to be assumed by medical professionals in other areas of primary care. There are doubts within the medical community about the adequacy of medical genetics education in medical schools for students not pursuing pediatrics or obstetrics. At the very least, primary care providers need to be equipped to discuss tests results and make necessary referrals. This requires a basic understanding of genetics. Yet, in a 1985 survey, only 21 percent of U.S. medical schools were considered to have good or excellent instruction in human genetics. Forty-seven percent of the schools responding were considered to have either nonexistent or poor human genetics teaching (27).

An OTA survey of companies developing predictive tests revealed that they had little confidence in the ability of primary care physicians to inform their patients about genetic screening, arrange for tests, and interpret test results (31). There is ample evidence that physicians have difficulty interpreting results of laboratory tests more familiar to them and less complicated than genetic tests (3,6,25).

Adequate genetic services are not always provided in the most likely setting-hospitals. In a study of Huntington's disease counseling in Veterans' Administration Hospitals, less than 1 percent of

the hospitals had a formal policy regarding the provision of genetic counseling (18).

While physicians are likely to be required to administer most genetic monitoring and screening tests, they are not the only health care professionals qualified to provide genetic services. Nurses, social workers, and master's level genetic counselors frequently participate in counseling and followup programs for individuals seeking genetic services. There are currently 15 programs in the United States and Canada offering a master's in genetic counseling (34). For many years, there has been some recognition that genetics is an important feature of the nursing curriculum (4) and, yet, when nurses have been surveyed about their genetic knowledge, important gaps have been noted (37). This has important implications for job site genetic monitoring and screening, as occupational health nurses are likely to be involved.

The American Board of Medical Genetics has certified more than 1,000 providers of genetic services, of which approximately half are clinical geneticists (M.D., D. O., or D.D.S.) (28). However, certification does not necessarily test one's counseling ability. Nonphysician genetic counseling personnel have been trained since 1969 and play a critical role in delivering services in an already overburdened system. One of the rate-limiting steps in the widespread use of genetic monitoring and screening tests will be the availability of adequately trained personnel to interpret results and provide followup services.

### *Cost of Counseling and Additional Tests*

***If an*** employer proceeds with monitoring or screening and then refers the worker to an outside source for additional testing or followup, unless the employer is willing to pay for those services, the costs of further testing or followup may deter some employees from proceeding. When tests are coupled with prenatal diagnosis or when multiple family members need to be evaluated for linkage studies, the bill can be well over $1,000. For example, the cost for predictive testing for Huntington's disease currently ranges from $2,800 to $4,000. This includes genetic and psychological counseling, a neurological examination, as well as posttest counseling (7).

Presently, some Blue Cross/Blue Shield Plans (BC/BS) and State Medicaid programs reimburse for genetic services, although services covered and amounts reimbursable vary. Reimbursement by Medicaid is frequently less than the full charge. This is particularly true for genetic counseling, which is sometimes not reimbursed at all (12). Fewer than half of BC/BS plans reimburse for carrier screening tests, and genetic counseling is covered by less than 60 percent (11). Twenty-three of the thirty-five health maintenance organization plans provide genetic counseling as a covered benefit.

Deficiencies in reimbursement for genetic counseling in both BC/BS and Medicaid programs are in part due to the absence of an American Medical Association code for genetic counseling (which is used by insurers to guide payment) and the policy of third-party insurers of not reimbursing nonphysician genetic counselors. Nonphysician genetic counselors are likely to be a needed source for referral of individuals identified through screening and monitoring programs. The fact that genetic consultations are frequently excluded in part, or in full, from insurance coverage is a disincentive for individuals pursuing further interpretation of their test results.

As part of the OTA survey on genetic monitoring and screening practices in the workplace, questions concerning genetic counseling were asked. The following section describes these results.

### *Use of Genetic Counseling: Survey Results*

Corporate health officers in companies (Fortune 500 and non-Fortune 500 companies) that have conducted any form of genetic monitoring or screening were asked:

> Has an employee ever been referred for genetic counseling by your company's medical staff as a result of any medical or genetic testing?

Health officers in 10 percent of those companies that had ever done genetic monitoring or screening reported that one or more employees in their companies had been referred to genetic counseling as a result of medical testing (table 8-l). Half of these companies were currently conducting some form of genetic monitoring or screening, and the other half had only tested in the past. Nearly all of the companies referring employees to genetic counselors (8 out of 9) had 10,000 or more employees.

OTA found that 6 percent of the companies that had conducted genetic monitoring or screening employed a genetic counselor. No companies re-

**Table 8-l-Genetic Counseling Referrals**

**Q. 26. Has an employee ever been referred for genetic counseling by your company's medical staff as a result of any medical or genetic testing?**

**(Base: Health Officers in companies that have ever done genetic monitoring or screening)**

| Unweighed base | Total percent (59) |
|---|---|
| Yes | 10 |
| NO | 80 |
| Don't know[a] | 5 |
| No answer | 5 |

a volunteered response.

SOURCE: Office of Technology Assessment, 1990.

ported contracting with a genetic counselor (table 8-2).

**Table 8-2 -Company Employment of Genetic Counselors**

**Q. 25. Does your company employ or contract with a genetic counselor?**

**(Base: Health Officers in companies that have ever done genetic monitoring or screening)**

| Unweighed base | Total percent (59) |
|---|---|
| Employ | 6 |
| Contract with | 0 |
| Neither | 89 |
| No answer | 5 |

SOURCE: Office of Technology Assessment, 1990.

## SUMMARY AND CONCLUSIONS

Individuals who have just learned about a genetic condition through employment genetic monitoring or screening face a double dilemma. Workers may have found that they are unemployable in certain job positions (including their current one) and that their future health or that of family members may be in jeopardy.

How individuals react depends on their own life circumstances as well as the diagnosis and prognosis. Because a probability, but not a certainty, that disease may result if difficult uncertainties are introduced into the lives of those tested. The information provided prior to administration of the test can help to prepare individuals for the outcome. In addition, a genetic counselor can help the person being tested understand the concept of risk. When the test results are positive, posttest counseling and followup are essential.

An important aspect of human communication is the context in which it occurs. Workplace testing is an atypical setting for receiving information of such personal importance. The absence of referrals to trained professionals and reimbursement for the costs of additional tests or counseling may be prohibitive factors influencing an individual's ability to obtain additional information. Current resources to provide counseling may be strained as more tests are developed and made commercially available.

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