Chapter 1

Summary, Policy Issues, and Options for Congressional Action

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Chapter 1

Summary, Policy Issues, and Options for Congressional Action

During the past three decades, our understanding of genetics has advanced remarkably as new methods for identifying, manipulating, and analyzing deoxyribonucleic acid (DNA) have developed. Less well understood, however, is the interaction between the environment and heredity, and the roles each plays in sickness and health. It has long been recognized that genetic risks are associated with certain workplace environments, such as exposure to radiation or certain chemicals. Recognition of genetic factors in disease presents new opportunities for detection, prevention, and treatment. This concept has provoked debate in recent years about whether genetic monitoring and screening of workers to identify outwardly healthy individuals (or populations) at risk for or susceptible to a variety of work-related conditions is appropriate or even feasible.

Genetic monitoring and screening have the potential to significantly change the workplace by detecting both occupational and nonoccupational diseases. They can identify genetic abnormalities which may be associated with inherited diseases, susceptibilities, and traits in otherwise healthy, asymptomatic individuals. The ability to diagnose latent conditions (both occupationally and nonoccupationally related) through genetic monitoring and screening raises policy questions about the proper use of such technologies.

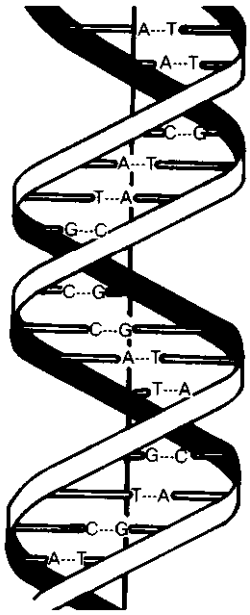
This report examines the potential applications and limitations of genetic monitoring and screening in the workplace. In response to requests from the Senate Committee on Commerce, Science, and Transportation, the House Committee on Energy and Commerce, and the House Committee on Science, Space, and Technology, this assessment presents the scientific, legal, ethical, and social issues surrounding the use of genetic monitoring and screening in the workplace. It also evaluates the results of a 1989 Office of Technology Assessment (OTA) survey on genetic monitoring and screening in the workplace of 1,500 U.S. companies, the 50 largest utilities, and the 33 largest unions. These survey results will also be interpreted in the context of a 1982 OTA survey on genetic monitoring and screening (part of a 1983 OTA assessment of genetic monitoring and screening).

DEFINING GENETIC TESTING

Genetic testing includes a number of technologies to detect genetic traits, changes in chromosomes, or changes in DNA. DNA is the chemical bearer of genetic information, which takes the structural form of a double-stranded helix (figure l-l). It is composed, in part, of four chemical subunits called bases. These four bases—guanine (G), adenine (A), thymine (T), and cytosine (C)---are the coding units of genetic information that form the DNA double helix structure (figure 1-2).

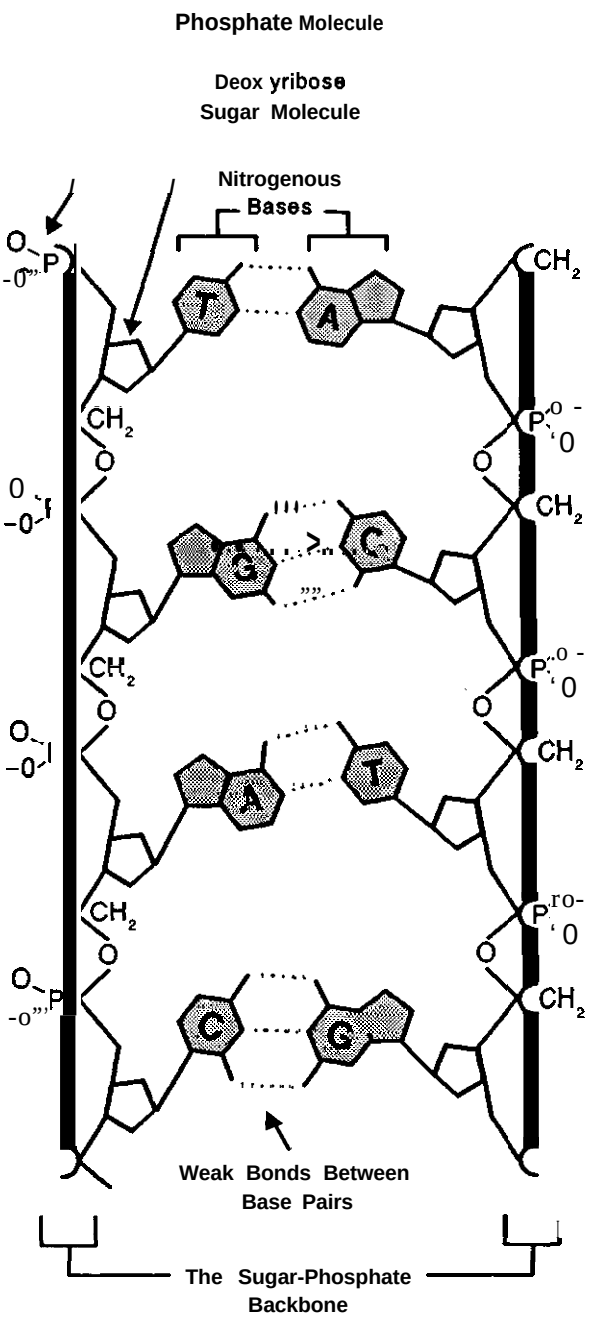
As used in the workplace, genetic testing encompasses two activities: genetic monitoring and genetic screening. Thus, genetic testing of employee populations involves both examining persons for evidence of induced change in their genetic material (monitoring) and identifying individuals with particular inherited traits or disorders (screening). The general term “genetic testing” is not used in this report; rather the more specific terms “genetic monitoring” and “genetic screening” are used (figure 1-3).

Figure l-l—The Structure of DNA



SOURCE: Office of Technology Assessment, 1990.

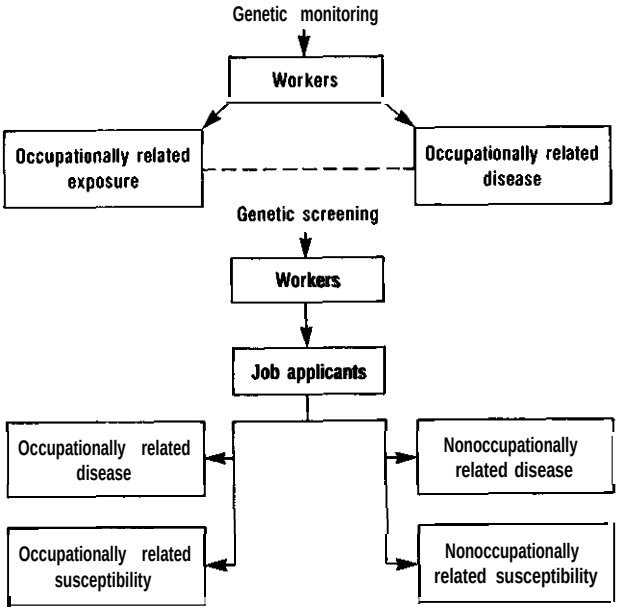
Figure 1-2—DNA Base Pairing



The four nitrogenous bases, adenine (A), guanine (G), cytosine (C), and thymine (T), form the four letters in the alphabet of the genetic code. The pairing of the four bases is A with T and G with C. The sequence of the bases along the sugar-phosphate backbone encodes the genetic information.

SOURCE: Office of Technology Assessment, 1990.

Figure 1-3-Components of Genetic Testing in the Workplace



SOURCE: Office of Technology Assessment, 1990.

What is Genetic Monitoring?

Genetic monitoring involves periodically examining employees to evaluate modifications of their genetic material--e.g., chromosomal damage or evidence of increased occurrence of molecular mutations—that might have evolved in the course of employment. The putative cause is workplace exposure to hazardous substances. The premise is that such changes could indicate increased risk of future illness.

Because ambient exposures, personal habits and lifestyle decisions (e.g., tobacco use, etc.), and age can also induce changes in genetic material, genetic monitoring could detect changes that arise from exposures outside of the workplace. In short, genetic monitoring ascertains whether the genetic material of a group of individuals has altered over time. In general, current techniques are not exposure-specific, but serve merely as an indicator of recent exposure.

Genetic monitoring could be performed on groups of employees to identify the risk for the exposed group as a whole, to target work areas for increased safety and health precautions, and to indicate a need to lower exposure levels for a group exposed to a previously unknown hazard.

What is Genetic Screening?

Genetic screening involves assays to examine the genetic makeup of employees or job applicants for certain inherited characteristics. (Employees could be screened on different occasions for different traits or with improved technology, but generally only once per characteristic.) Genetic screening can be used in two distinct ways. First, employees or job applicants could be screened for the presence of genetically determined traits that render them susceptible to a pathological effect if exposed to specific agents. For example, an employee or a job applicant could be tested to identify a genetic predisposition to an occupationally related disease. Second, employees or job applicants could be screened to detect general heritable conditions, not just conditions associated with occupational illness. Reasons for using the different classes of tests vary. In either case, whether screening for an occupationally related trait or one unrelated to job exposure, genetic screening tests involve examinations for inherited traits where a single measure is usually sufficient because these inherited characteristics, as a rule, do not change.

Genetic screening for occupationally related traits could be performed to ensure appropriate worksite placement of employees susceptible to certain occupational diseases, and ensure that employers place those workers most susceptible to a specific risk in the least hazardous environments. Both genetic screening for occupationally related traits and for nonoccupationally related traits could be performed to: improve employee productivity and lower workers' compensation costs through better worker health; promote and encourage general health awareness; and improve employers' health care cost-containment efforts, especially for health insurance. This could be done through exclusion (i.e., not hiring those with deleterious genes because of the potential drain on health insurance).

Genetic screening differs significantly from genetic monitoring. With screening, a one-time test to detect a single trait in a worker or job applicant is usually sufficient, while monitoring generally involves multiple tests of a worker over time. Most importantly, genetic screening focuses on the preexisting genetic makeup that workers or job applicants bring to the job. This is distinct from genetic monitoring which focuses on hazardous workplace exposures that induce changes in the genetic material in an exposed population as a whole.

DIFFERENCES BETWEEN GENETIC MONITORING AND SCREENING

From a policy standpoint, these differences—genetic monitoring v. genetic screening and occupational illness v. nonoccupational illness or general health--could be significant. Some criticize all types of genetic monitoring and screening in the workplace as paternalistic and discriminatory, while others advocate that, properly implemented, genetic monitoring and screening programs benefit both workers and industry. Others, however, maintain that it is one thing to monitor or screen workers because they are at increased risk for occupational illness induced by the workplace, but quite another to screen persons because they or their offspring—who could be covered on an employee's health plan—are at high risk for a disease unrelated to occupational exposure. Finally, some argue that genetic screening *per se*, even if to reduce occupational illness, is unfair because it ***a priori*** measures heritable conditions beyond an individual's control. Genetic monitoring is perceived by others as less threatening because it mirrors other forms of successful biological monitoring (e.g., benzene or lead exposure) performed on body fluids or tissue samples. The use of either technology, however, raises serious legal and ethical questions.

Screening for nonoccupationally related diseases-e. g., Huntington's disease or neurofibromatosis—raises new issues for containing health care expenses, for both the employer and employee. Increasingly, costs to U.S. employers of health-related benefits have skyrocketed. In particular, to avoid rising health care costs, many large companies are adopting self-insurance plans, which are not subject to State insurance regulation. Self-insurance refers to the practice of employers, particularly large employers, assuming the risks for the health care expenses of their employees instead of purchasing health insurance through insurance companies. Companies concerned about health insurance costs could be interested in screening workers and job applicants who are likely to develop genetically based diseases and could impose high costs on a company's self-insured health program. Similarly, companies could engage in genetic monitoring—

again, to safeguard workers' health while simultaneously reducing the burden of occupational illness on their health care costs. Corporate "wellness" programs, or other company-sponsored health promotion programs (box l-A) that emphasize prevention and encourage employees to adopt healthier lifestyles, are one way companies can limit their health care expenses.

In addition to the obvious and significant benefits from preventing serious illnesses, genetic monitoring and screening for occupationally related disease could provide indirect benefits, such as a reduction in the costs associated with occupational illnesses for employees and their families, employers, the insurance industry, and society. Workplace health risks can produce financial costs to the worker in the form of medical bills, changes in insurance status, lost wages, ill health, and, in some cases, premature death. When a worker incurs an occupational illness, the employer experiences lower productivity levels, higher insurance premiums, workers' compensation claims, and potential lawsuits. Insurance companies either sustain a loss or raise others' premiums. And, society pays for the care and compensation of some with occupational illnesses through Federal health programs. The magnitude and distribution among the sectors of society of the benefits and costs of genetic monitoring and screening will help determine the desirability of this approach to improving occupational health.

HISTORY OF GENETIC MONITORING AND SCREENING

The concept of genetic monitoring and screening surfaced before the discovery of DNA by Watson and Crick in 1953. As early as 1938, noted geneticist J.B.S. Haldane introduced the idea of sorting workers according to their susceptibilities. One of the first cases of an individual's genetic condition reacting to either a chemical agent or drug was reported in the 1950s, when some American soldiers in Korea experienced hemolysis (the destruction of red blood cells). The hemolysis was attributed to their carrier status of glucose-6-phosphate dehydrogenase (G-6-PD) deficiency, which results in less of the enzyme G-6-PD in their red blood cells. It was later postulated that carriers of G-6-PD deficiency could also undergo hemolysis after exposure to certain chemicals. The possibility of conducting a preplacement examination to detect employees with the trait was considered.

In the 1970s, there was considerable public interest in nonoccupational screening programs for sickle cell anemia. These programs became the focus of controversy and criticism because proper genetic counseling was not always provided, and results were not always kept confidential. As a result, discrimination sometimes occurred in the workplace, and from insurance companies. (Federal and State legislation in this area is discussed in a later section.)

Incidents of industry involvement in genetic monitoring or screening since the 1960s have been reported. They have varied from research programs using genetic monitoring techniques for evaluating chromosomal damage to efforts in genetic screening to detect conditions such as G-6-PD deficiency or sickle cell trait.

GENETIC MONITORING AND SCREENING IN THE WORKPLACE: A HISTORY OF CONGRESSIONAL CONCERN

Congressional interest in human genetics, genetic diseases, and genetic technologies is not new. In 1972, Congress passed the National Sickle Cell Anemia Control Act (Public Law 92-294), amending it 4 years later to the National Sickle Cell Anemia, Cooley's Anemia, Tay-Sachs, and Genetic Diseases Act (Public Law 94-278). The goals of both pieces of legislation included increased levels of basic and applied research, training, testing, counseling, and public education in the area of screening for sickle cell anemia and other genetic diseases. More recently, congressional interest in human genetics has focused on the mapping and sequencing of the human genome.

The 1983 Office of Technology Assessment Report

In the late 1970s and early 1980s, reports surfacing about genetic monitoring and screening in occupational settings captured the interest of Congress. Concern about scientific and social issues of such testing prompted the House Committee on Science and Technology to hold hearings and request an OTA assessment of *The Role of Genetic Testing in the Prevention of Occupational Disease.*

Box 1-A+ancer Detection in the Workplace

Among the greatest fears of industrial workers is the risk of cancer from exposure to hazardous substances. Although employees are concerned about cancer risk, they are not always informed about the specific dangers of the chemicals with which they work. By increasing employee and employer involvement m cancer prevention and detection, both groups stand to benefit: employees with gains m personal health, and employers with higher worker morale and productivity and reduced health expenditures. Because cancer risks vary from worksite to worksite, worker perceptions of various job hazards related to cancer and chemical exposure are important. From 1978 through 1987, the National Cancer Institute (NCI) allocated $14 million to the Occupational Safety and Health Administration (CM-IA) for cancer prevention training and education of workers. In 1983, NCI awarded grants to five unions, that had participated in OSHA's education program, to evaluate the impact of the unions' cancer prevention and education programs.

A 1987 study by the International United Rubber, Cork, Linoleum, and Plastic Workers of America, ***one*** of the participating unions, questioned approximately 24,000 of its members about their knowledge of chemical hazards, the location of engineering controls, and the usc of daily safety procedures. Prior to the study, employees had participated in the industry's cancer control program, which included worker education.

Despite the fact that over 10,000 different chemicals, many hazardous, are used by these workers, the study found that 22 percent of workers were not sure whether they worked with dangerous chemicals, and only 6 percent felt they were very informed about chemical hazards. Percentages of employees saying they were well-informed varied widely from company to company, ranging from 16 percent at one company to 32 percent at another. Thus, for adequate cancer education, greater understanding of chemical-specific risks is needed.

In addition to worker perception and involvement, management health programs can play an important role. Currently, several companies offer employees cancer screening clinics and other cancer detection programs. One such program, offered by Pennzoil (in conjunction with the Kelsey -Seybold Foundation) to employees at a Texas facility, began in 1984 as a cancer awareness clinic for white-collar employees to discover cancers unrelated to worksite exposure. The Pennzoil program, strictly voluntary and confidential, has since been expanded to industrial petroleum workers and other locations in 22 States, where the cancer detection procedure can include workplace risks.

The Pennzoil program involves an initial lecture on cancer risk and detection, and a personal cancer examination for those requesting one. Corporate management strongly supports these meetings, and encourages employees to attend the lectures. As part of the program, employees also complete questionnaires about cancer risk behaviors and personal medical histories. Those employees showing an increased risk of cancer are offered followup counseling sessions with Kelsey-Seybold Foundation Cancer Prevention Center physicians and medical tests, if necessary, paid for by Pennzoil. AU employees also are offered yearly screening or followup examinations. Pennzoil receives only summary data on participation, cancer detection, and demographic information. Both increased employee morale and detection of potential tumors resulted Along with Pennzoil's expansion of the program, Exxon Chemical Americas has undertaken a similar project with the Kelsey-Seybold Foundation Cancer Prevention Center.

The experience of cancer screening in the workplace suggests that the cooperation of corporate management and private organizations, together with an accurate assessment of employee understanding of workplace risk can create a healthier, more productive working environment; one benefiting both employer and employee. At present, genetic monitoring detects genetic changes that could result in greater risk of cancer. Future advances in genetic technologies could result in increased cancer testing and education at worksites. As genetic technologies make detection of cancer or other health risks more accurate, programs such as those just described could seine as models. For genetic monitoring and screening in particular, implementing successful worker education will be crucial.

SOURCES: Office of Technology Assessment, 1990, based on M. Minkoff, Keky-Seybold Foundation Cancer Prevention Center, Houston, TX, personal communication, October 1988; A.P. Schenck, A.D. Kaluzny, G.M. Hochbaum, et al., "Worker Perceptions and Actions Toward Cancer Control in the Workplace: An Analysis of Baseline Data, " and L.. Zimmerman, G. G. Jackson, J. Hughes, et al., ''Cancer Education and Screening in the Workplace: The Corporate Perspective, Advances in *Cancer Control: The War on Cancer--l5 Years of Progress,* P.F. Engstrom, L.E. Mortenson, and P.N. Anderson (eds.) (New York, NY: Alan R. Liss, Inc., 1987).

As part of its study, OTA surveyed American industry and unions to determine the extent and nature of employer genetic monitoring and screening (the 1982 OTA survey results are compared to the 1989 survey results in a later section).

In the intervening years, several developments have led to renewed congressional interest in assessing the current extent of and issues surrounding genetic monitoring and screening in the workplace. Understanding of human molecular genetics and biotechnologies applicable to the field have expanded enormously. Both the technical capability to detect genetically based disorders and the number of applications of such technologies have increased. Finally, the use of other types of employee testing (e.g., acquired immunodeficiency syndrome (AIDS), drug, and polygraph) as well as the current efforts to map the human genome, also combined to stimulate congressional interest.

Impacts of the Human Genome Project

Efforts underway to map and sequence the human genome stand to have a significant impact on many aspects of biology, medicine, and health-including genetic monitoring and screening. To date, genome projects have accelerated the production of new technologies, research tools, and basic knowledge. At current or perhaps increased levels of effort, they may eventually make possible the control of many human diseases-first through more effective methods of predicting or detecting disease, and ultimately, in some cases, through development of effective therapies based on improved understanding of disease mechanisms. Although not a direct result of the genome project, advances in human genetics and molecular biology have already provided insight into the origins of such diseases as cystic fibrosis, hemophilia, sickle cell disease, and hypercholesterolemia.

The new technologies developed through human genome projects research will also be used to assess public health needs. Techniques for rapidly sequencing DNA, for example, may facilitate the detection of mutations following exposure to radiation or environmental agents. Susceptibilities to environmental and workplace toxicants might be identified as more detailed genetic linkage maps are developed. In addition, special methods of surveillance could be used to monitor individuals at risk.

However, possible applications of and access to these genetic data pose profound ethical questions. The complexity and urgency of these issues will increase in proportion to advances in mapping and sequencing. The human genome project will certainly accelerate diagnostic applications. Progress to date indicates that the ability to diagnose a genetic abnormality precedes the development of therapeutic interventions and that this gap may be growing. Access to this information by third-parties (e.g., insurance companies or employers) and how this information is used are important related issues. These questions are complex and are unlikely to be resolved in the near future. As a means to explore these issues a working group on ethics was established in January 1989 by the Program Advisory Committee on the Human Genome. Additionally, a percentage of the Federal genome budget will go toward studying the ethical issues associated with the genome research.

THE STATE-OF-THE-ART

In 1983, OTA found that none of the genetic tests evaluated met established scientific criteria for routine use in an occupational setting. However, OTA determined that enough suggestive evidence existed to merit further research. Since that time, rapid progress in both human molecular genetics and occupational medicine has increased understanding of causal relationships between disease and environmental factors, including workplace exposure to a variety of substances. This report reexamines the technologies available and evaluated by OTA in 1983 in light of new developments and knowledge in this area. In addition, this report more thoroughly evaluates the area of genetic screening for nonoccupationally related traits and diseases. Finally, it also examines novel techniques (e.g., restriction fragment length polymorphisms (RFLPs)) and tests (e.g., Huntington's disease).

Genetic Monitoring Technologies

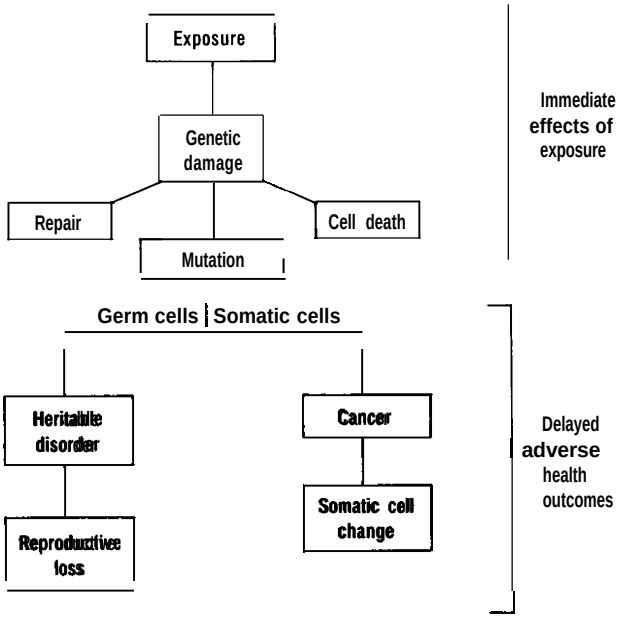
Genetic monitoring ascertains whether an individual's genetic material has altered over time. Workplace genetic monitoring is designed to detect the effects of a toxic substance or its byproducts, and to evaluate the genetic damage caused by such a substance. The objective of these techniques, ultimately, is to predict risk of disease due to genetic damage. When hazards are identified via genetic monitoring, prevention programs can be considered

that will reduce exposures to hazards. This is of particular concern for certain occupational groups that are exposed to such hazardous substances over many years at much higher concentrations than the general population.

It is well-documented that exposure to some chemical substances and to radiation at high doses causes cancer and genetic mutations (changes in genetic information). Not all mutations, however, cause disease (figure 1-4). The relationships between genes, mutations, and disease are becoming clearer with the development of molecular techniques. However, until the health effects of radiation and chemical exposures are better understood, genetic and biological monitoring of exposed populations can only provide a gross indication that genetic changes have occurred and that adverse health effects could follow. Changes in a cell's genetic material (DNA) can be detected at either the chromosomal level, using cytogenetic methods which detect major structural changes in chromosomes, or at the molecular level using noncytogenetic methods.

The application of cytogenetic tests to measure chromosomal damage is based on the concept that

Figure 1-4-Biological Consequences of Exposure to Mutagenic Agents



SOURCE: office of Technology Assessment, adapted from J.B. Ward, 'Issues in Monitoring Population Exposures," *Carcinogens and Mutagens in the Environment Volume //, The* Workplace, Hans F. Stich (cd.) (Boca Raton, FL: CRC Press, 19S5).

Photo credit: U.S. Council for Energy Awareness

A Nuclear Power Worker: Controversy continues regarding the carcinogenic effects of radiation in employees of the nuclear weapons and nuclear power industries.

damage to cells' genetic material represents initial events in a process that may eventually lead to disease. Cytogenetic methods can detect human exposures at biologically significant levels in populations, but the interpretation of findings for the individual remain uncertain. In some cases, detectable mutations result from gross changes in chromosome structure and can be visualized under the microscope. The disruptive effects of mutagens on chromosome structure, organization, and behavior have long been studied by geneticists. However, the connections between chromosomal damage and disease are unclear except in a small number of cancer cases. Most analysts agree that interpretation of cytogenetic results at the individual level is questionable and recommend that until the relationship between cytogenetic damage and disease is better understood, interpretation should be limited to the population level. In addition, cytogenetic monitoring of human populations is expensive and time-consuming. There can also be technical variations associated with both test limitations and interpretations.

Until recently, most tests for mutagenicity have been merely indicators of exposure, only providing evidence that exposure has occurred. This limitation is diminishing with the development of more techniques at the molecular level, thus refining the ability to document exposure and, in some cases, providing qualitative information. New molecular assays of mutagenicity, e.g., hypoxanthine-guanine phosphoribosyltransferase and oncogene protein detection, are providing greater specificity and will

augment tests already in use, e.g., the Ames test. New methods may provide better estimates of the health effects of low doses of some mutagens, as well as providing qualitative data on the nature of mutation. Detecting activated oncogenes and DNA adducts has the potential of predicting disease in asymptomatic individuals. As the nature of mutation becomes more clearly defined, the connection between mutation and disease will also become better understood.

A genetic monitoring or screening test must be proved valid and reliable before a decision can be made on its value. Validity is the probability that a test will correctly classify true "positive" and true ''negative' results. Tests of the same specimen must repeatedly give the same result whether performed by several different laboratories or by the same laboratory on several occasions to be reliable. If the tests are valid and reliable, establishing procedural safeguards and designing well-conceptualized test protocols can avert erroneous and misleading conclusions. The use of genetic monitoring methods in epidemiologic studies will continue to be plagued by problems associated with classical approaches to determining hazardous exposures in the workplace. Eliminating biases, obtaining controls, and keeping good records are procedural difficulties that may be encountered. The employment of more specific and sensitive tests, rather than the reliance on any one test for valid and reliable results, will lead us closer to understanding the relationships between exposure, mutation, and disease.

Genetic Screening Technologies

It has long been speculated that genetically determined variation in susceptibility may predispose some workers to occupational disease while others in the same environment seem to be unaffected. Genetic influences may be exaggerated or diminished by one's age, diet, or overall health status. Recognition of genetic factors in disease (both occupationally and nonoccupationally related disease) presents new opportunities for detection, prevention, and treatment.

In assessing the state-of-the-art in screening tests for use at worksites, three different questions must be discussed:

- What general techniques are presently available that could be used for genetic screening?
- What is known about the association among heritable traits, exposure to hazardous materials, and subsequent occupational illness?
- What genetic disorders unrelated to job exposures that are important to general health can be detected?

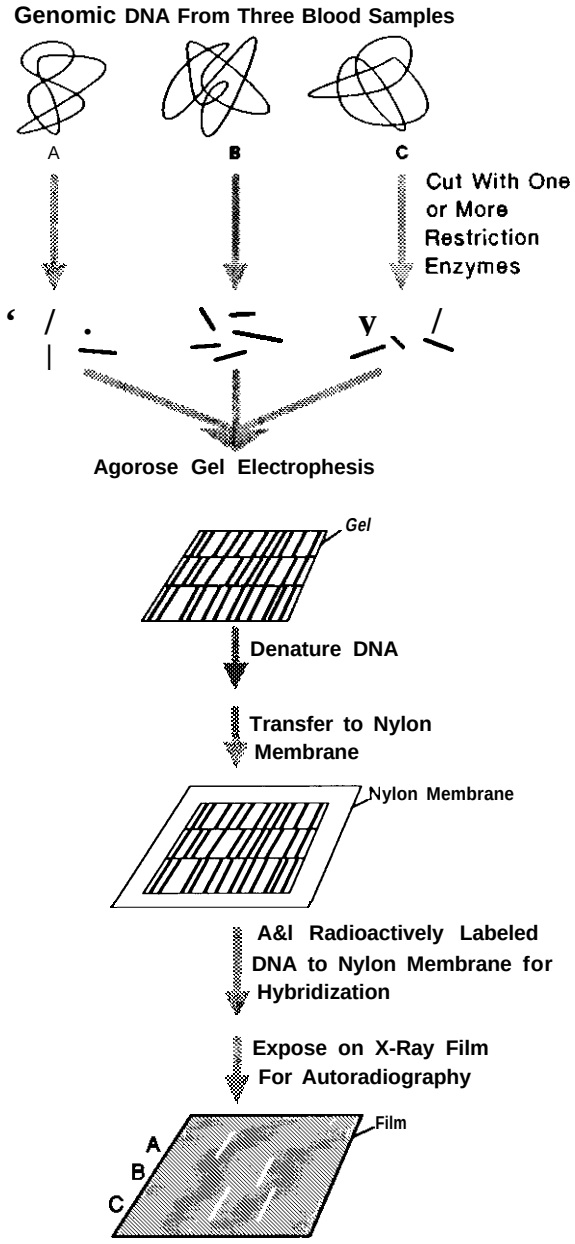
Since the 1983 OTA report, there have been several technical advances in genetic screening tests. In addition, several new susceptibilities to occupational disease have been identified, and progress in detecting some nonoccupationally related disorders has been made. These nonoccupationally related disorders, which are likely to affect large populations, might be of interest to an employer if they can be detected through preemployment screening.

Biochemical and molecular techniques for detecting genetic disease are discussed in this report. Biochemical genetics refers to the analysis of mutant genes on the basis of altered proteins or metabolizes. If diagnosed, some of these "inborn errors of metabolism" can be treated with enzyme replacement or dietary control. An example of such a biochemical disorder is phenylketonuria, which can be controlled by restricting dietary intake of the amino acid phenylalanine. In general, biochemical techniques for diagnosing genetic disease are often restricted to indirect analysis of gene products rather than diagnosis targeted at the gene itself.

Advances in DNA technology have greatly enhanced our ability to directly examine the genetic basis for disease and to predict and diagnose such diseases in larger populations. Until recently, most available tests for genetic conditions were not based on recombinant DNA techniques. Today, DNA-based tests encompass a variety of standard diagnostic techniques that allow examination of regions very near the genes (e.g., Huntington's disease) or direct examination of the genes themselves (e.g., sickle cell anemia and cystic fibrosis).

Two important tools, RFLPs, which serve as markers for the presence of a diseased gene, and cloned DNA probes, represent the major advances responsible for improved diagnosis of genetic disease (figure 1-5). Another technology, polymerase chain reaction (PCR) has also facilitated the ability to detect genetic disorders. PCR can be thought of as molecular photocopying (figure 1-6). PCR itself is not used directly to analyze DNA, but allows a scientist to take a sample that ordinarily would be insufficient to detect the characteristics of the DNA,

Figure 1-5-Detection of Restriction Fragment Length Polymorphisms Using Radioactively Labeled DNA Probes



Variations in DNA sequences at particular marker sites are observed as differences in numbers and sizes of DNA fragments among samples taken from different individuals (shown here as samples A, B, and C).
SOURCE: Office of Technology Assessment, 1990.

and reproduce it until enough DNA copies are available for examination by a number of technologies, including RFLP analysis. Chapter 5 discusses RFLP analysis and PCR in greater detail.

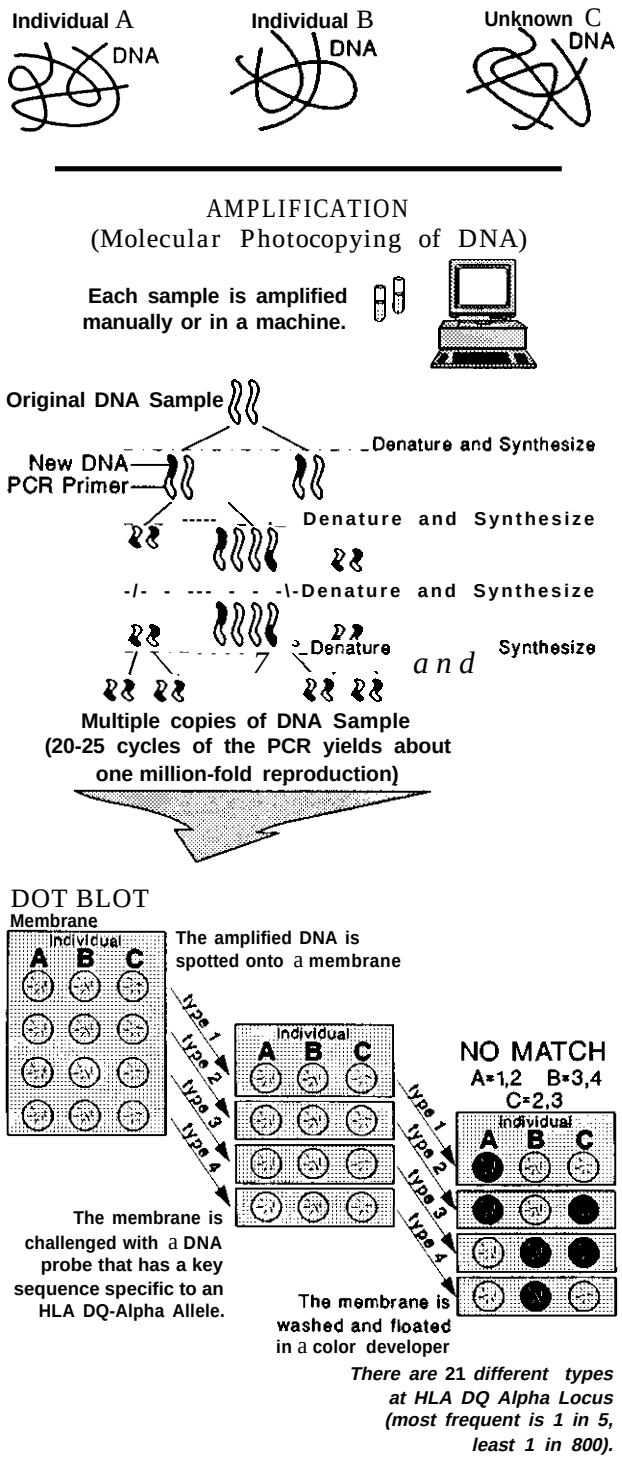
The obstacles to understanding associations between predisposition and disease are slowly eroding as the use of synthetic probes, PCR, and automated DNA-sequencing machines increase the efficiency and lower the cost of mass screening. However, before widespread screening of populations is begun, the validity of the tests should be determined. Also, quality control is likely to become a major issue as the volume of tests performed at laboratories grows. These are already issues in forensic applications of DNA-based tests.

At present, there are approximately 50 diseases that have the potential to enhance an individual's susceptibility to the toxic or carcinogenic effects of environmental agents. These occupationally related diseases include: G-6-PD deficiency, sickle cell trait, and the thalassemias (see table 1-1 for more examples).

Molecular biology has enhanced the traditional determination of "predisposition to disease' (previously based on physical examination, family history, and lifestyle habits) by seeking out and finding genes or markers associated with disease. Individuals found to have the gene or the marker can then be identified, sometimes with near certainty, to be candidates for disease. Often, predisposition only manifests in disease when there is an accompanying environmental insult, e.g. toxic substances, viruses, or other disease. The influence of the environment, however, remains the wild card in most cases, because possession of the genetic predisposition alone may be insufficient to cause disease. It is likely that for some time modern science will be more successful in identifying the genes and the markers than in identifying the environmental agent(s) necessary for activation of the predisposing genes.

Predispositions to certain cancers have been the focus of much research in the past few years. As the associations between carcinogenesis and genetics become clearer, the boundaries between occupational and genetic disease may become more blurred. (Box 1-B describes some of the connections between genetic damage and cancer.) Research on the predispositions to atherosclerosis, diabetes, mental illness, and chemical addiction has also progressed in recent years. In addition, research is providing insight into possible genetic predispositions to such common ailments as lower back injuries, obesity, allergies, and arthritis. While

Figure 1-6-The Polymerase Chain Reaction



SOURCE: Office of Technology Assessment, 1990.

predictive tests are not immediately foreseeable in any of those areas, as more populations are studied and more linkage maps prepared, it is possible that screening tests will be developed.

With accelerating interest in tests to detect abroad range of genetic disorders and increasing investment in biotechnology industries, the market demand for tests, especially DNA-probe tests, is expected to expand. While the population affected by genetic conditions for which tests are available is still somewhat small, the potential future test population for multifactorial diseases is enormous (see table 1-2).

WHAT LEGAL CONSIDERATIONS ARE INVOLVED?

Existing legal concepts must strain to keep pace with the scientific advances of genetic monitoring and screening. Only a limited body of law dealing directly with genetic monitoring and screening in the workplace exists. There is, however, a substantial body of law pertaining to the related practice of medical testing of workers, which may influence the legal issues associated with genetic monitoring and screening in the workplace.

Because an individual's genetic makeup is not chosen, legal questions of the most sensitive sort are raised in seeking to analyze these personal characteristics. Genetic monitoring and screening raise legal questions related to workplace safety and employee rights. Among the fundamental legal issues arising from genetic monitoring and screening in the workplace are:

- privacy from unwanted monitoring or screening;
- confidentiality of the information obtained;
- potential discrimination in employment opportunities; and
- ultimately, the health of the subject.

OTA examined common and statutory law—both of which have a bearing on genetic monitoring and screening in the workplace. Changes in the common law relating to workplace genetic monitoring and screening have been incremental over recent years. An increasing body of case law is developing, however, over employer screening for drug use and AIDS.

Table I-I-Identification and Quantification of Genetic Factors Affecting Susceptibility to Environmental Agents

High-risk groups	Estimated occurrence	Environmental agents to which group is (may be) at increased risk
RBC conditions		
G-6-PD deficiency	American Black males 16%; Mediterranean Jewish males 119'.; Greeks 17.-2%; Sardinians 1%-8%	Environmental oxidants such as ozone, nitrogen dioxide, and chlorite
Sickle cell trait	7Y0-13Y0 of American Blacks are heterozygotes	Aromatic amino and nitro compounds; carbon monoxide, cyanide
The thalassemias	Alpha: 4%-5% in Americans of Italian and Greek descent; Beta: 2%-7% American Blacks and 2%-3% American Greeks	Lead; benzene
NADH dehydrogenase deficiency (MetHb reductase deficiency)	Estimated 1% of population are heterozygotes	MetHb-forming substances
Catalase Hypocatalasemia	About 2% of U.S. population based on Swiss gene frequency	Ozone; radiation
Acatalasemia	1/10,000-20,000 of U.S. population based on Swiss gene frequency	
Low SOD activity	Frequency of genetic variants in population 1 to 2/10,000; normal population exhibits unimodal distribution; persons at low end of distribution may beat increased risk	Wide variety of environmental oxidants; paraquat; radiation; ozone
ALA dehydratase deficiency	Unknown, but thought to be rare	Lead
Hb M	Unknown, but rare	Carbon monoxide
Erythrocyte potphyria	1.5/100,000 in Sweden, Denmark, Ireland, West Australia; 3/1 ,000 in South African Whites; rare in Blacks	Chloroquine; hexacholorobenzene; lead; various drugs, including barbiturates, sulfonamides, others
GHS-Px deficiency	Rare	Environmental oxidants
GSH deficiency	Rare	Environmental oxidants
Liver metabolism		
Defect in gluocuronidation		
Gilbert's syndrome	6% of normal, healthy adult population	Wide variety of xenobiotics including polychlorinated biphenyls
Crigler-Najjar syndrome	Few persons live to adulthood	
Defect in sulfation	Unknown	Wide variety of xenobiotics; best association is with tyramine-containing foods
Acetylation phenotype, slow v. fast	Slow: 50% Whites; 50% Blacks; 10% Japanese Fast: 50% Whites; 50% Blacks; 90% Japanese	Aromatic amine-induced cancer; numerous drugs, e.g., isoniazid and hepatitis
Gout	0.27%-0 .3%. prevalence in U.S. and Europe	Lead
Oxidation center defects	9% of British Whites; 8% of Nigerians; 6% Ghanians; 1%. Saudi and Egyptians are poor oxidizers	Numerous xenobiotics requiring oxidative metabolism for detoxification
OCT deficiency	Unknown, but thought to be rare	Insect repellant (DET)
Paraxonase variant	25%-30% of population	Parathion
Rhodanese variant	Unknown	Cyanide
Sulfite oxidase deficiency heterozygotes	Unknown	Sulfite, bisulfite, sulfur dioxide
Inadequate carbon disulfide metabolism	Upward Of 30%-40%	Carbon disulfide
Alcohol dehydrogenase variant	5%. English; 20% Swiss; 70% Japanese	Metabolize (e.g., ethanol) more quickly than normal
Wilson's disease	Homozygous 1/100,000 while the heterozygote may approach 1/500	Cooper, vanadium
Serum variants		
Albumin variants	Less than 1/1 ,000 in Europeans, much higher frequency in North American and Mexican Indians	Unknown
Pseudocholinesterase variants	Highly sensitive homozygous and heterozygous persons of European ancestry have combined frequency of about 1/1,250; moderately sensitive genotypic variants of European ancestry have frequen cy of 1/1 5,000	Organophosphate and carbamate insecticides; muscle relaxant drugs

* Abbreviations used are: G-6-PD, glucose-6-phosphate dehydrogenase; NADH, nicotinamide adenine dinucleotide (reduced form); SOD, superoxide dismutase; ALA, aminolevulinic acid: GSH, reduced glutathione; Px, peroxidase; OCT, ornithine carbamoyl transferase; IgA, immunoglobulin A; PKU, phenylketonuria; PTL, phenylthiourea; XP, xeroderma pigmentosum; AT, ataxia telangiectasia; FA, Fanconi's syndrome.

SOURCES: E.J. Calabrese, *Ecogenetics: Genetic Variation in Susceptibility to Environmental Agents* (New York, NY: Wiley Interscience, 1984); E.J. Calabrese, "Ecogenetics: Historical Foundation and Current Status," *Journal of Occupational Medicine,* 28(10):1096-1102, 1986.

Box 1-B--Genetics and Cancer

Cancer is a genetic disease arising from genetic damage of diverse sorts—recessive and dominant mutations, large rearrangements of DNA and point mutations--all leading to distortions of either the expression or biochemical function of genes. The growing field of cancer genetics aims to uncover the genetic alterations responsible for uncontrolled growth of cancer cells. Many types of human cancer occur in familial as well as sporadic forms. Discrete genetic changes have been associated with different types of neoplasm, and are thought to initiate or cause progression of cancer. Chromosome studies in more than 10,000 cases of neoplasm have reported specific anomalies. The identification of genetic changes, therefore, presents the major diagnostic challenge in cancer.

Both dominant and recessive forms of cancer have been found. The genetics of the common cancers--breast, colon, and lung-are beginning to fit a pattern, Approximately 5 percent of cancer cases constitute a hereditary cancer syndrome in which a dominant gene predisposes to cancers of the breast, ovary, brain, gastrointestinal system, and white blood cell precursors. These are referred to as "cancer families' (see ch. 5 for further discussion). Apart from these, each type of cancer appears to have a small group of cases that fits the pattern of a major predisposing gene and a much larger group that seems to be largely environmenta1 in origin.

In addition, examples such as xeroderma pigmentosa imply that there is a connection between susceptibility to cancer and impaired ability of cells to repair damaged DNA. It is a reasonable expectation that if cancer is related to alterations in somatic cell genes, then the rate at which those changes occur could serve as a barometer of changes in the gem-dine that may not be expressed for many generations to come.

Certain cancers, e.g., lung, laryngeal, bladder, and testicular, have repeatedly been linked to environmental exposures. The effects on chromosomes of such chemicals as arsenic, asbestos, chromium, nickel, and vinyl chloride are well-documented. Substances that cause chromosomal abnormalities are called ''clastogens. The reader is referred to the 1983 Office of Technology Assessment report for more detail on the specific effects of those agents. Chapter 5 describes recent advances in detecting predisposition to some common cancers.

SOURCES: Office of Technology Assessment, 1990, based on J.M. Bishop, "The Molecular Genetics of Cancer, " *Science* 235:305-311, 1987; F. Mitelman and J.D. **Rowley, "Genes,** Chromosomes and Cancer: A New Forum for Research in Cancer Genetics," Genes, *Chromosomes & Cancer* 1:1-2, 1989; J.J. Nora and F.C. Fraser, *Medical Genetics: Principles and Practice* (Philadelphia, PA: *Lea* & Febiger, 1989); R. Parshad, K.K. Sanford, *K.H. Kraemer,* et al., ' 'Carrier Detection in Xeroderma Pigmentosum," *The Journal of Clinical Investigation* 85: 135-138, 1990; U.S. Congress, Office of Technology Assessment, *The Role of Genetic Testing in the Prevention of Occupational Disease,* OTA-BA- 194 (Washington, DC: U.S. Government Printing office, April 1983).

Of particular interest in terms of common law developments is the apparent continuing expansion of the public policy exception to the employment at-will doctrine for dismissal from employment. This rule forms the basis for most employment relationships, absent an explicit contract between the parties, and gives the employer virtually unlimited authority to terminate the employment relationship at any time. It includes the right to refuse to hire an individual because of a perceived physical inability to perform the job and the right to terminate employment because of a belief that the employee is no longer able to perform adequately. With respect to genetic monitoring and screening, this would allow an employer to use either in any way, including personnel decisions. Even if test results were inaccurate or unreliable, the employer would be protected in basing employment actions on them. In recent years, however, courts have begun to erode the scope of the at-will doctrine by creating exceptions. While exceptions to employment-at-will have grown to cover different grounds for dismissal, they have not been applied to other kinds of employment actions. This trend may also play an important role in forming judicial attitudes toward employment decisions based on genetic monitoring and screening results.

Beyond the role of occupational health and safety regulation, common law decisions regarding confidentiality and privacy are relevant to genetic monitoring and screening in the workplace. An area of concern is the role of the occupational health physician in genetic monitoring and screening. Because the occupational health physician is hired by the employer, there can be some question whether legal precedents protecting confidentiality in the physician-patient relationship apply, Occupational health physicians must balance patient privacy and confidentiality on the one hand with employer need-to-know on the other.

Table 1-2—Genetic Tests Available and Total Americans Affected

Genetic condition	Total cases
Currently ***available:***	
Adult polycystic kidney disease	500,000
Fragile X Syndrome	100,000
Sickle cell anemia	65,000
Duchenne muscular dystrophy	32,000
Cystic fibrosis	30,000
Huntington's disease	25,000
Hemophilia	20,000
Phenylketonuria	16,000
Retinoblastoma	10,000
Total	798,000
Potential future tests:	
Hypertension	58,000,000
Dyslexia	15,000,000
Atherosclerosis	6,700,000
Cancer	5,000,000
Manic-depressive illness	2,000,000
Schizophrenia	1,500,000
Type 1 diabetes	1,000,000
Familial Alzheimer's	250,000
Multiple sclerosis	250,000
Myotonic muscular dystrophy	100,000
Total	89,800,000

SOURCE: *Medical World News,* p.58,Apr. II,1988.

Photo credit: Margaret Anderson

U.S. Department of Labor, Washington, DC: Headquarters of the Occupational Safety and Health Administration.

Federal Regulatory Framework

Federal legislation ranging from the Occupational Safety and Health Act (OSH Act) (Public Law 91-596), the Rehabilitation Act of 1973 (29 U.S.C. 791 et seq.), Title VII of the Civil Rights Act of 1964 (42 U.S.C. 2000e), the National Labor Relations Act (NLRA) (29U.S.C. 151 et seq.), and the Americans with Disabilities Act (ADA) (Public Law 101-336) provide some protections against abuses, particularly those relating to unilateral employer imposition of genetic monitoring and screening, discrimination based on monitoring and screening results, and breaches of confidentiality concerning results.

The OSH Act contains several federally imposed statutory duties related to occupational safety and health that must be carried out by the employer. The Occupational Safety and Health Administration (OSHA) has regulated some employer practices that could have a bearing on genetic monitoring and screening, including employee access to medical records and communications about hazards by employers to employees.

Title VII of the Civil Rights Act of 1964 prohibits discrimination in employment practices based on race, color, religion, sex, or national origin. In addition to intentionally discriminatory actions, neutral employment practices that have a disparate impact on a protected group may violate Title VII. A Supreme Court decision, *Wards Cove v. Atonio,* recently placed stringent standards on workers attempting to use statistics to prove discriminatory employment practices. This decision could make it more difficult for an employee to prove that an employer's employment practices are discriminatory. Many genetic screening procedures (e.g., sickle cell disease, G-6-PD deficiency) have a disparate impact that could fall under the protection of Title VII.

The Rehabilitation Act of 1973 bans discrimination against handicapped individuals by employers who are government contractors or recipients of Federal assistance. In order to fall under its protection, an employee must prove that his or her genetic trait is or is regarded as an impairment, and in addition, he or she must otherwise be qualified. Accordingly, an individual with a genetic predisposition for a disease may not be denied employment or promotion simply because of the predisposition so long as the individual is otherwise qualified for the position. In such a case, the

Photo credit: Earl Dotter

The use of protective clothing can help prevent occupational illness and injury.

employer would have to make reasonable accommodation for the person.

ADA, which was recently signed into law and whose enforcing regulations have yet to be drafted and approved, extends a clear and comprehensive prohibition of discrimination on the basis of disability to the private sector. It bans discrimination against individuals with disabilities in hiring, discharge, compensation, or any term, condition, or privilege of employment by an employer engaged in an industry affecting commerce. By 1994, this legislation will apply to all employers with 15 or more employees. Whether a genetic marker or a trait constitutes an 'impairment' under ADA is unclear. Preemployment medical examinations or inquiries are to be used only as a tool for determining the applicant's ability to do the job. Thus, genetic screening for nonoccupationally related conditions would seem to be prohibited. ADA language, however, does not specifically address genetic monitoring or screening.

NLRA governs the relationships of employees, labor organizations (unions), and employers engaged in businesses affecting interstate commerce. Safety and health matters, including fitness-for-duty physical examinations and medical testing are considered mandatory subjects of bargaining between these parties. Thus, genetic monitoring and screening could be considered mandatory subjects of collective bargaining. In this context however, preemployment screening of job applicants would not be covered. Additionally, not all employees are union members and therefore would not be covered under collective bargaining agreements.

The protections provided by current Federal legislation are somewhat disjointed, applying at times to applicants and at times to workers, or offering protections of which applicants and workers may be unaware. The exact role of each will depend on the nature of the tests that are developed and their application. While it is clear that many legal tools presently exist, it is probable that new ones will be needed as unexpected challenges arise.

On the whole, it appears that Federal regulatory law, as administered by OSHA, is likely to have the most immediate impact on the use of genetic monitoring and screening in the workplace. OSHA has dealt extensively with related practices of biological monitoring that could forma ready source of rules for genetic monitoring and screening. OSHA's rules on access to medical records and hazard communication are among the most directly applicable sources of existing law. Thus, OSHA is the most appropriate candidate for regulating in the area of genetic monitoring and screening in the workplace. It could call on the National Institute for Occupational Safety and Health (NIOSH) to provide research and recommendations for regulatory development. However, at this time the OSH Act neither prohibits nor requires genetic monitoring or screening.

State Responsibility

States have a role in a variety of areas concerning genetic monitoring and screening. One of these areas is workers' compensation programs which are designed to provide no-fault compensation to workers suffering harm as a result of their employment. A particular challenge is posed, however, by diseases with long latency periods between exposure to a causative agent and onset of illness. It is the compensation of such long latency conditions that presents the most likely opportunity for the application of genetic monitoring and screening. Many issues concerning genetic monitoring and screening and workers' compensation (e.g., the role of genetic

data as evidence and their admissibility, and the coverage of a susceptible employee) are unresolved.

Some States have directly addressed genetic discrimination. OTA found in 1983 that four States had passed statutes limiting the use of genetic information in employment decisions. In three of these States—Florida, Louisiana, and North Carolina-the laws are specific to testing for sickle cell trait. In New Jersey, however, a fairly broad measure was passed banning employment discrimination based on genetic traits. If this measure becomes a model for other jurisdictions, the adverse impact, and perhaps benefits, of genetic monitoring and screening results on employees could be severely curtailed. The New Jersey experience will be interesting to observe as more genetic monitoring and screening tests become available.

WHAT ETHICAL ISSUES ARE INVOLVED?

Genetic monitoring and screening involve the acquisition of personal information in the workplace. Several important ethical issues arise when discussing the interests of employees, job applicants, employers, and society, including the potential for discrimination. Many of these issues express the same concerns as current laws: legal and ethical arguments often share common ground.

Yet, while legal and ethical issues can be similar, approaches to resolve dilemmas raised differ. Law does not reflect all moral values held by members of society, nor can it necessarily be used to resolve ethical dilemmas. Ethical arguments about the use of genetic monitoring and screening in the workplace often address obligations, rights, or values not explicitly covered by law. Awareness of the ethical issues surrounding new technology is essential for formulating and implementing policies that reflect the greatest possible regard for human values.

Although the ethical issues show little change since the 1983 OTA study, the emphasis placed on some concerns about genetic monitoring and screening has shifted. In 1983, OTA found that genetic monitoring and screening were not inherently unethical, and that if they were used to enhance worker health in a reamer consistent with ethical principles, they could be morally justified. Whether the tests were consistent with ethical principles depended on how they were done and how the resulting information was used. Since that time, there has been increased pessimism in public debate about the risks genetic screening for nonoccupationally related disease could have for employees' autonomy and privacy. Attention has shifted from the uncertain technical efficacy of genetic monitoring and screening in predicting or identifying illness to the potential abuses of genetic monitoring and screening in the workplace.

At least three parties play a role and have an interest in genetic monitoring and screening in the workplace--job applicants and workers, employers, and society. For these parties, three principal issues exist:

- the implementation of genetic monitoring and screening tests in the workplace and the use of the information they generate;
- the dissemination and storage of information gained from genetic monitoring and screening; and
- the role of genetic counseling for both employers and employees in genetic monitoring and screening programs.

Each of these issues is probably viewed differently by job applicants and employees, employers, and society, since each group has different interests to protect. In addition, the ethical issues associated with genetic monitoring and screening in the workplace vary according to whether the test performed is genetic monitoring for chromosomal damage, genetic screening for susceptibilities to occupational illness, or genetic screening for inherited conditions or traits unrelated to the workplace.

Employees and job applicants, for example, want to protect their autonomy and privacy. They could feel that all genetic information should remain confidential under any circumstance, especially if it might be used to deprive them of a job, health insurance, or other benefit (box l-C). Employers, in desiring to preserve their liberty to make their own hiring decisions, might want to be free to conduct monitoring or screening programs. They might also wish to establish the conditions for employee participation and consequences for those who refuse to participate. Such practice would be consistent with current preemployment medical testing practices. Society has an interest in promoting a safe workplace, and fair treatment of individuals, as well as economic efficiency.

Box 1-C-An International Survey of Attitudes of Medical Geneticists Toward Workplace Genetic Screening and Access to Results

A survey on mass genetic screening was sent to 1,053 medical geneticists in 18 nations, of whom 677 responded. Geneticists strongly preferred voluntary over mandatory workplace screening, by a 72 percent majority. In the United States, there was consensus (greater than 75 percent) that screening should be voluntary. Geneticists who thought screening should be voluntary cited the worker's autonomy or right to decide (74 percent), and the danger of stigmatization, discrimination in employment, or misuse of information by institutional third-parties (41 percent). Advocates of mandatory screening cited protecting the individual worker's health (64 percent), protecting public health (51 percent), and efficiency or cost-benefit arguments (22 percent). Nine percent of those who advocated voluntary screening and 12 percent of those who advocated mandatory screening based their responses in part on concern for economic interests of employers.

In advocating voluntary versus mandatory screening, a clear difference of opinion on whose welfare the respondent placed foremost was reported. Ninety-seven percent who advocated voluntary screening and 58 percent who advocated mandatory screening placed the worker's welfare as most important. Three percent who advocated voluntary screening and 37 percent who believed in mandatory screening placed societal interests first. Only 1 percent placed the employer's welfare first.

Advocates of voluntary screening were more likely than supporters of mandatory screening to describe a conflict of interest between worker and employer, 34 percent described such conflicts, as opposed to 13 percent who advocated mandatory screening. A majority of both groups, however, described no conflicts.

Ninety-eight percent of respondents said the worker should have access to test results, including 86 percent who said the worker should be told the results even if he or she did not ask for them. When asked whether the employer should have access to genetic screening results, 81 percent said employers should have no access without the worker's consent, including 22 percent who believed that employers should have no access at all.

Thirty percent of respondents who gave reasons for their choices about access believed it would be to the worker's benefit if the employer had some form of access, e.g., employers could shift susceptible workers to less dangerous jobs. Only 6 percent of respondents, however, thought that working conditions m general would be improved. Nineteen percent described potential economic disc rimination, stigmatization, or other misuse of test results by employers. Ten percent based their responses on the economic interests of the employer.

When asked whether government health departments should have access, 68 percent said there should be no access without worker consent. There was a strong consensus on this issue in six nations, including the United states.

These differences in perception indicate that geneticists-those in a position to conduct genetic screening tests-share concerns about how genetic screening might be used in the workplace. A large majority of geneticists ranked the interests of workers and society above those of employers in importance, but they disagree about how those interests can be best protected. Many geneticists believe that workplace screening should be voluntary and that workers should make autonomous decisions about whether to undergo genetic screening. Almost all geneticists believed workers should receive genetic screening results, but that employers' access should be restricted 'hey expressed concern that genetic screening results may be used to justify personnel actions that may stigmatize or discrimina te against some workers.

SOURCE: D.C. Wertz and J.C. Fletcher, "An International Survey of Attitudes of Medical Geneticists Toward Mass Screening and Access to Results," *Public Health Reports* 104(1):35-44, 1989.

A balance must be struck between promoting one party's autonomy and compromising that of another. If employers are free to implement and enforce genetic monitoring or screening policies, the autonomy of job applicants and employees will be limited. Conversely, giving the applicant or employee complete freedom to protect his or her own interests would restrict the freedom of the employer and, in some instances, present risk to co-workers or family.

Employer and employee interests can conflict at three points in the processes of genetic monitoring and screening:

- the decision to undergo genetic monitoring or screening;
- access to information gained from genetic monitoring or screening; and
- the communication and interpretation of genetic monitoring or screening results.

Certain broad guidelines for the use of genetic monitoring and screening could at least partially address the concerns of all parties. Such guidelines could produce maximal benefits to all parties—minimizing occupational illness without threatening privacy or confidentiality, denying equality of opportunity, or stigmatizing workers.

GENETIC COUNSELING FOR INDIVIDUAL USES OF GENETIC INFORMATION

The effects and results of genetic monitoring or screening transcend the workplace, and raise issues for the individual who is tested-not just as a worker—but as a person and family member. As a result of new technical capabilities to diagnose and predict genetically based disease, pathways for informed decisionmaking about ourselves and our family's health have expanded. However, these capabilities often create moral, ethical, and psychological dilemmas for which no easy solutions exist. Receiving such personal information in the workplace setting differs from the way most people learn about their genetic identity, because the individual may not have sought to be tested. When tests are conducted in a medical setting, a context is provided in which certain assumptions and expectations can reasonably be held by the person being tested. These factors might be different when the workplace becomes the background for receiving genetic information.

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, 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. 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. 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.

However, doubts can be introduced into the lives of those tested because genetic monitoring and screening tests often convey a probability, but not a certainty, that disease will appear. When it is not possible to give an accurate recurrence risk or more than a general diagnosis, the interactions between the test subject and the test administrator are even more complex. In the case of genetic monitoring, it is likely that nonspecificity of diagnosis and prognosis will predominate. Further complicating the use of monitoring and screening tests is the fact that for most genetic diseases, effective

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.

Box 1-D--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 asymtomatic 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 choose to be tested, even though there is a 50 percent chance that they will receive good news Prior to the availability of a predictive teat 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 screend, counseled, and provided followup care. In addition, it suggests that 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 Presymtomatic 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," Journal of the ***American Medical Association*** **257:3362 (letter),** 1987; B. Teltacher and S. Polgar, "Objective Knowledge About Huntington's Disease and Attitudes Toward Predictive Tests of Persons at Risk," *Journal of Medical 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 Genetics 20:179-188, 1983.**

interventions are not yet feasible (box l-D). Employers undertaking genetic monitoring and screening programs should anticipate the complexity of interpretation and communication of test results.

The workplace is an atypical setting for receiving information of such personal importance. It should not be overlooked that when genetic monitoring or screening are used in the workplace, the focus of the tests--the person—is being provided with information that could have a significant impact on decisions unrelated to employment: marriage, procreation, and lifestyle. 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. Genetic counseling and appropriate referrals for those at risk should accompany the use of either genetic monitoring or screening.

SURVEY OF THE USE OF GENETIC MONITORING AND SCREENING

To assess the current practice of genetic monitoring and screening by U.S. employers, a survey was conducted for OTA from March 24 to July 15, **1989,** by Schulman, Ronca, & Bucuvalas, Inc. This effort is a followup to a **1982 survey that was part of the 1983** OTA report. As with the earlier survey, the core remained a national survey of the ***500*** largest U.S. industries (Fortune **500), 50 largest utilities, and 33 major unions. The 1989 survey was designed** to provide comparability to the earlier survey in terms of populations sampled and the questionnaire content. The **1989 survey, however, did not exactly duplicate the 1982 questionnaire. Rather, it was designed to remove ambiguities that might have been present in the initial survey, but could be detected only in hindsight. It also was designed to include a representative sample of all other compa-**

Table 1-3-Summary of Methodology

Samples	
Fortune 500 companies . .	Sampled in 1989 and 1982.
50 largest utilities	Sampled in 1989 and 1982.
Unions	33 unions in 1989 and 11 unions in 1982.
Companies with 1,000+ employees	1,000 sampled in 1989. Not sampled in 1982.
Designated respondent	
Private companies:	
Chief health officer	Designated respondent in 1989 and 1982. Received version of questionnaire for health officers.
Chief personnel officer. .	Designated respondent in 1989 only. Received different questionnaire version for personnel officers.
Unions:	
Union president	Designated respondent in 1989 and 1982.
Followup methodology	
Reminder letters	Sent in 1989 and 1982.
Remailing questionnaires to nonresponders	Sent to all nonresponders in 1989 and 1982.
Telephone followup to nonresponders	All Fortune 500 and utilities in 1989. Only 200 largest companies in 1982.
Actual telephone interviews with nonresponders to mail survey	Done as a last resort in 1989 and 1982.

SOURCE: Office of Technology Assessment, 1990.

nies with 1,000 or more employees so that broader estimates could be made of the use and pattern of genetic monitoring and screening in the workplace. (See table 1-3 for a summary of the methodology of the 1989 and 1982 surveys.)

To flesh out the details of the data from the 1989 survey, OTA added questions that explored the use of genetic monitoring and screening in greater depth. Questions were asked about genetic monitoring and screening tests that might have been conducted as part of a voluntary wellness program, at the request of the employee, or for diagnosis. Including the results of the new questions produced a broader definition of genetic monitoring and screening for the 1989 survey. OTA believes the increased specificity attained an accurate measure of genetic monitoring and screening in 1989, established a firm base for future comparisons, and preserved general comparability to the 1982 study.

Table 1-4-Current Use of Genetic Monitoring by Fortune 500 Companies

Q.15. Is your company currently conducting *cytogenetic monitoring* of any employees or job applicants, for research or any other reason?

Q.18. Is your company currently conducting *direct-DNA monitoring* of any employees or job applicants, for research or any other reason?

(Base: Health officers)

	Number of companies currently conducting	
	Cytogenetic monitoring	Direct-DNA monitoring
Total	1	0
Type of business		
Electrical utility	0	0
Pharmaceutical	0	0
Other chemical	0	0
Petroleum.	1	0
Electronic	O	0
Other manufacturing	0	0
Nonmanufacturing	0	0
Number of employees		
Less than 5,000	O	0
5,000-9,999	0	0
10,000 or more	1	0

SOURCE: Office of Technology Assessment, 1990.

Table 1-5--Current Use of Genetic Screening by Fortune 500 Companies

Q.13. Is your company currently conducting *biochemical genetic screening of any* employees or job applicants, for research or any other reason?

Q.17. Is your company currently conducting *direct-DNA screening of* any employees or job applicants, for research or any other reason?

(Base: Health officers)

	Number of companies currently conducting	
	Biochemical genetic screening	Direct-DNA screening
Total	12	0
Type of business		
Electrical utility	0	0
Pharmaceutical	0	0
Other chemical	4	0
Petroleum	1	0
Electronic	O	0
Other manufacturing	2	0
Nonmanufacturing	5	0
Number of employees		
LOSS than 5,000	1	0
5,000-9,999	2	0
10,000 or more	9	0

SOURCE: Office of Technology Assessment, 1990.

Before presenting the trend data from 1982 to 1989, the current, past, and combined use of genetic monitoring and screening will be discussed. Following those sections, the overall use of genetic

Table 1-6-Past Use of Genetic Monitoring Tests by Fortune 500 Companies

Q.16. Has your company conducted any cytogenetic *monitoring* of any employees or job applicants, for research or any other reason in the past 19 years?
(Base: Health officers)

	Number of companies conduoced in past
	Cytogenic monitoring
Total	5
Type of business	
Electrical utility	0
Pharmaceutical	0
Other chemical	1
Petroleum	0
Electronic	0
Other manufacturing	1
Nonmanufacturing	3
Number of employees	
Less than 5,000	0
5,000-9,999	0
10,000 or more	5

SOURCE: Office of Technology Assessment, 1990.

Table 1-7—Past Use of Genetic Screening Tests by Fortune 500 Companies

Q.14. Has your company conducted any biochemical*genetic* screening of any employees or job applicants, for research or any other reason in the past 19 years?
(Base: Health officers)

	Number of companies conducted in past
	Biochemical genetic screening*
Total	8
Type of business	*o*
Electrical utility	
Pharmaceutical	0
Other chemical	4
Petroleum	0
Electronic	0
Other manufacturing	2
Nonmanufacturing	2
Number of employees	
Less than 5,000	0
5,000-9,999	1
10,000 or more	7

NOTE: Includes companies currently conducting genetic screening.
SOURCE: Office of Technology Assessment, 1990.

Table 1-8-Combined Testing: Current v. Past Monitoring and Screening by Fortune 500 Companies

(Base: Health officers)

	Number of companies
Conducted genetic monitoring or screening for research or any other reason, at present or in past 19 years	20
Currently conducting genetic monitoring or screening	12
Conducted monitoring or screening in past only	8
Conducted genetic screening for research or any other reason at present or in past 19 years	16
Currently conducting genetic screening	12
Conducted genetic screening in past only	4
Conducted cytogenetic monitoring for research or any other reason at present or in past 19 years	6
Currently conducting cytogenetic monitoring	1
Conducted cytogenetic monitoring in past only	5
Currently conducting direct-DNA screening for research or any other reason	0
Currently conducting direct-DNA monitoring for research or any other reason	0

SOURCE: Office of Technology Assessment, 1990.

monitoring and screening in 1989 and 1982 will be discussed.

In the 1982 survey, six health officers (1.6 percent) reported their companies currently conducted genetic monitoring or screening. In 1989, 12 health officers (5 percent) reported their companies currently conducted genetic monitoring or screening (1 of the companies used genetic monitoring and screening while 11 used only genetic screening) (see tables 1-4 and 1-5). (These numbers do not directly correlate because of different sized survey populations in 1982 and 1989.) The increase in the number of "current users' in 1989 could reflect slight differences in question wording between the 1989 and 1982 surveys.

The 1989 survey asked health officers whether their companies had conducted genetic monitoring tests in the past 19 years, for research or any other reason. Five health officers in Fortune 500 companies reported that their companies had conducted cytogenetic monitoring in the past 19 years of any employees or job applicants, for research or any other reason (table 1-6). All five companies that formerly conducted cytogenetic monitoring reported no current use of genetic monitoring or screening. A total of eight health officers in the Fortune 500 companies reported that their companies had conducted biochemical genetic screening of any employees or job applicants in the past 19 years (table 1-7). This included four health officers in Fortune 500 companies that reported they were currently conducting biochemical genetic screening.

A total of 20 health officers reported that their companies had conducted cytogenetic monitoring or

Table 1-9-Use of Genetic Monitoring or Screening: 1989 v. 1982 Survey Results

(Base: Health officers)

	Number of companies	
	1989	1982
Conducted genetic monitoring or screening for research or any other reason, at present or in the past*	*20*	*18*
Currently conducting genetic monitoring or screening	12	6
Conducted monitoring or screening in past only	8	12

● Defined as past 19 years in 1989 survey and past 12 years in 1982 survey.

SOURCE: Office of Technology Assessment, 1990.

Table I-10-Consideration To Conduct Genetic Monitoring and Screening in the Next Five Years: Health Officers

Q.33. Does your company anticipate conducting any *biochemical genetic screening,* for any reason, in the next 5 years?

Q.34. Does your company anticipate conducting any *cytogenetic monitoring,* for any reason, in the next 5 years?

Q.35. Does your company anticipate conducting any *direct-DNA screening,* for any reason, in the next 5 years?

Q.36. Does your company anticipate conducting any *direct-DNA monitoring,* for any reason, in the next 5 years?

(Base: Health officers)

	Percent			
	Yes	No	Not sure	No answer
Biochemical genetic screening	4	218	25	3
Cytogenetic monitoring	1	219	27	3
Direct-DNA screening	0	224	23	3
Direct-DNA monitoring	1	218	27	4

SOURCE: Office of Technology Assessment, 1990.

biochemical genetic screening, either currently or in the past 19 years. This includes 12 health officers who reported that genetic monitoring or screening was currently conducted, and 8 who reported that genetic monitoring or screening had been conducted in the past 19 years, but not currently (table 1-8). (In the 1982 OTA survey, past was defined as 12 years, and in the 1989 survey, as 19 years.)

Trend data on the use of genetic monitoring or screening can be obtained by tabulating comparable questions in the 1989 and 1982 surveys. These do not include the previously mentioned items added in 1989. Using this narrow definition, of the 330 companies (62.4 percent) responding to the 1989 survey, 20 health officers reported that their companies had conducted genetic monitoring or screening, either currently or in the past 19 years. In comparison, the 1982 survey found 18 health officers in the Fortune 500 sample who reported current or past use of genetic monitoring or screening (table 1-9). Thus, there has been little change between 1989 and 1982 in the number of companies that had used genetic monitoring or screening in the workplace.

In summary, the 1989 survey found 12 companies reporting current use of genetic monitoring or screening for research or any other reason. The ratio of current to past use of monitoring or screening was reversed in 1982, with 6 companies indicating current use of genetic monitoring or screening and 12 companies indicating past but not current use. Overall, OTA found that 20 companies had used genetic monitoring or screening in 1989, as compared to 18 companies in 1982.

If there has been little or no growth in the number of companies conducting genetic monitoring and screening in the workplace, what do companies foresee for the future? In 1982,, OTA found that 4 companies (1. 1 percent) anticipated using the tests in

the next 5 years, and 55 companies (15 percent) stated they would "possibly" use the tests in the next 5 years. The 1989 OTA survey provided the response categories "yes," "no," and "not sure" for the same questions to avoid classifying an indefinite answer as a positive response to future genetic monitoring or screening.

OTA found one Fortune 500 company that anticipated cytogenetic monitoring, one company that anticipated direct-DNA monitoring, and four companies that anticipated biochemical genetic screening. No company anticipated using direct-DNA screening in the next 5 years. Twenty-seven companies in 1989 indicated they were not sure whether they anticipated cytogenetic monitoring, and 27 were not sure whether they anticipated direct-DNA monitoring. For biochemical genetic screening, 25 companies were not sure whether they anticipated using it, and 23 were not sure about future direct-DNA screening (table 1-lO).[1] In 1982, 55 companies said they would possibly use such test in the next 5 years. Although this number cannot be directly compared to the current survey, the 1989 OTA survey appears to indicate fewer companies anticipate using genetic monitoring or screening.

POLICY ISSUES AND OPTIONS FOR CONGRESSIONAL ACTION

While technologies associated with genetic monitoring and screening in the workplace have continued to advance, OTA found no significant change in the use of these technologies since 1983. Thus, several of the policy issues and options for congressional action offered in the 1983 OTA report are still valid and remain unchanged.

Two central issues related to genetic monitoring and screening in the workplace were identified during the course of this assessment. They are:

- the appropriate role of the Federal Government in the regulation, oversight, or promotion of genetic tests (both monitoring and screening); and
- the adequacy of federally sponsored research on the relationships between genes and the environment.

Associated with each policy issue are several options for congressional action, ranging in each case from taking no specific steps to making major changes. Some of the options involve direct legislative action. Others involve the executive branch but with congressional oversight or direction. The order in which the options are presented does not imply their priority. Moreover, the options are not generally mutually exclusive; adopting one does not necessarily disqualify others that pertain to the same or other issues, although changes in one area could have repercussions in others. A careful combination of options might produce the most desirable effects.

ISSUE: Is there a role for the Federal Government in ***genetic monitoring*** in the workplace?

Option 1: Take no action.

Congress could take no action to prohibit, regulate, or promote the use of genetic monitoring in the workplace. This would allow employers, employees, and their representative trade groups and unions to regulate its use through negotiation, arbitration, and litigation.

Thus far, executive agencies involved in workplace health and safety have not regulated against the use of genetic monitoring in workplace settings. OSHA has regulated some employer practices that could affect the use of genetic monitoring, such as medical records access by the employee. Congress could take no action if it determines that present Federal regulation is adequate in this area. Under this scenario, constraints on the use of genetic monitoring would develop through court rulings in suits between pa-ties or by negotiations between companies and unions.

Option 2: Prohibit genetic monitoring in the workplace.

To prevent all possibilities for discrimination and breach of confidentiality, Congress could prohibit genetic monitoring in the workplace. In light of the many discrete changes needed in the OSH Act, NLRA, and Rehabilitation Act to achieve this degree of protection through regulation, Congress could decide to prohibit all genetic monitoring until further research into the methods is conducted. Such a

[1]These numbers cannot be added because of cross counting; nor do they directly correlate to the 55 companies because of question wording. In retrospect, those who chose "possibly' in 1982 might not have meant to indicate that genetic monitoring or screening was anticipated, they simply could not rule out the possibility they would use it in the future.

prohibition could shift the focus of the issue to levels of exposure in the workplace.

Prohibiting genetic monitoring, however, will delay the accumulation of data needed to make the judgment whether certain genetic monitoring tests are useful. By slowing the development of these data, prohibition might threaten efforts to identify workplace hazards, whether they are to be minimized through cleanup or worker selection. In addition, some workers who might have avoided dangerous exposures had they known of a susceptibility will sicken unnecessarily. Finally, this option clearly eliminates the possibility for mandatory worker protection under those discrete circumstances where overall worksite hazard reduction is not technologically or economically feasible.

Option 3: Promote genetic monitoring in the workplace.

Congress could decide that genetic monitoring in the workplace should be promoted because of its potential to improve the work environment and worker health conditions. This could be done by providing additional funding to those Federal agencies currently performing research into genetic monitoring methods, as well as basic research on the cause of occupational disease, in general, and the relationships between environmental exposures and health effects arising from genetic mutation. Such projects could identify useful occupational genetic monitoring tests and develop protocols for their use. However, many questions about the use of genetic monitoring remain unanswered. Because the interpretation of genetic monitoring is only considered to be reliable at the population level, rather than the individual level, the current usefulness of genetic monitoring in the workplace is questionable.

ISSUE: Is there a role for the Federal Government in ***genetic screening*** in the workplace?

Option 1: Take no action.

Congress could choose to take no action in the area of genetic screening in the workplace. As with genetic monitoring, any constraints on the use of genetic screening would develop through court rulings in lawsuits between employers and employees, or by negotiations between companies and unions. In support of this option is the viewpoint that congressional action is not currently warranted at this time. Use of genetic screening in the workplace has not changed greatly since the 1983 OTA report. However, there have been several newly recognized susceptibilities to occupational illness since that time. In addition, advances have been made in the area of molecular techniques for genetic screening for both occupationally and nonoccupationally related disease.

If Congress takes no action in this area, those identified as susceptible to *occupational illness* through genetic screening could be seen as unfit for work. In addition, those identified as being susceptible to a *nonoccupationally related disease* could be seen as a health insurance burden. Without proper restrictions, the use of genetic screening to detect either type of disease risk could make job discrimination a possibility.

Option 2: Prohibit genetic screening in the workplace.

The principal reason for prohibiting genetic screening in the workplace would be the concern over its potential misuse. Such potential for misuse probably would be greater for genetic screening than genetic monitoring because the former is targeted toward identifying *individuals* **at increased** risk while the latter focuses on *groups* at increased risk. The existing legal framework may offer protection in some circumstances, but many questions have not resolved.

A drawback to this option is that by prohibiting both types of genetic screening in the workplace, employers could not utilize screening for occupationally related disease. This type of screening offers some protection of the worker.

Option 3: Promote genetic screening in the workplace.

Congress could stimulate the research and development of genetic screening tools by providing funds for research into occupationally related disease, nonoccupationally related disease, or both. Useful screening tests for occupationally related disease could be developed which could have direct benefits for the individual worker and indirect benefits for the employer. In addition, more money could be provided in the area of research on occupationally related traits. NIOSH could be authorized to do research in this area, and to certify procedures for medical technologies that are of sufficient value to be used in an occupational setting. If such research were promoted, however, employers might be prone to use those screening tests to screen

out susceptible employees in lieu of cleaning up the workplace.

If research in the area of nonoccupationally related disease was promoted beyond current Federal research levels, employers might be prone to using such screening tests to discriminate against employees or job applicants who might increase the company health care costs.

ISSUE: Should the Federal Government regulate genetic monitoring or screening in the workplace?

If Congress determines that the current regulatory framework addressing genetic monitoring and screening is adequate, it could take no action and let the current regulatory framework stand. However, if Congress determines that the current regulatory framework is inadequate, it could pursue several avenues. A framework established by several major pieces of legislation exists on which to build: OSH Act; NLRA; Title VII of the Civil Rights Act of 1964; Rehabilitation Act of 1973; and ADA. The following options are discussed according to whether they apply to genetic monitoring, genetic screening, or both.

Genetic Monitoring and Screening

Option 1: Congress could amend Section 6(b)(7) of the OSH Act which states that OSHA standards shall prescribe the type and frequency of medical examinations or other tests to be made available, to specify whether genetic monitoring and screening tests are to be included.

To either prohibit or promote genetic monitoring and screening, Congress could amend this section's coverage with respect to genetic monitoring and screening. To contain abuses, Congress could include language directing OSHA to prescribe or recommend genetic monitoring or screening only when less intrusive medical tests will not provide information of substantially the same value. Thus, for example, tests for sickle cell anemia would not be permitted unless other tests of lung fiction and blood oxygenation were incapable of giving an employer the information needed to decide whether a particular worker could safely manage a particular task.

A principal drawback of this option is that performing genetic monitoring and screening tests on employees could be financially prohibitive for some employers. In addition, mandating genetic monitoring or screening tests could be burdensome for both the employer and the employee. Such an action could require the employer to hire new medical staff to perform them. Furthermore, the employee might not wish to undergo genetic monitoring or screening.

Option 2: Congress could amend the OSH Act to guarantee the confidentiality of genetic monitoring and screening results.

Congress could amend the OSH Act to specifically guarantee that genetic monitoring and screening results not be disseminated, except in nonidentifying, statistical forms for research purposes, to any third-party without specific authorization from the worker. Further, employers could receive only the conclusion of the occupational physician, i.e., whether the worker is fit for the job in question, without receiving details or results of the genetic monitoring or screening tests. The worker, on the other hand, would receive both the test results and the conclusions drawn from them by the examining physician. Several State statutes provide a model for such legislation.

Advantages of this option are the ability to shield workers from misuse of genetic information by immediate and potential employers, and the maintenance of adequate authority to provide statistical information needed for ongoing improvement of health and safety practices. As with the option just mentioned concerning recordkeeping, however, this amendment would logically be appropriate to all medical records, and not merely those concerning genetic monitoring and screening tests. Thus, evaluating this option requires a larger consideration of whether the OSH Act should guarantee the confidentiality of all medical testing in this fashion. If this option is adopted, consideration would also need to be given to remedies for breach of confidentiality and an examination of the role of the occupational physician employed by the company.

Option 3: Require full disclosure to employees and job applicants of the nature and purpose of all medical procedures performed on them.

Current law does not require employers to disclose the nature and purpose of medical procedures conducted on employees or job applicants, or how the results are to be used. Although employees are given access to their medical records, they may not

be able to interpret the data within the records, or challenge incorrect information. A congressionally mandated requirement that employers provide detailed information of what procedures were performed and why they were performed might serve as a deterrent to abuses. This would also protect the employees' autonomy by allowing them to be part of a decisionmaking process that affects their health and economic interests. If the test were genetic in nature, the assistance of a genetic counselor would be important to fully explain the procedure and the meaning of a positive result.

On the other hand, by requiring full disclosure, Congress would place requirements on employers that might be perceived as burdensome and expensive. Additionally, arguments might be made that such a requirement would intrude on the judgment of the occupational health physician.

Genetic Monitoring

Option 4: Congress could direct OSHA to clarify that genetic changes shall be included under the definition of occupational illness.

OSHA's definition of occupational illness now includes "abnormal condition,' but does not specifically cover genetic changes. Taking this action could ensure that data on worker exposures and subsequent genetic changes would be recorded in worksites where employers are using genetic monitoring. This would help with ongoing efforts to assess the effects of potentially hazardous substances, as well as offer the opportunity to more closely monitor the health of a particular worker.

Yet including genetic changes in the definition of occupational illness would implicitly equate all genetic changes with "illness." Many changes are likely to be without immediate symptomatic effect. Therefore, gathering and distributing this information might be unduly alarming, particularly to the workers in question. Also, all genetic change is not definitely a result of the workplace. Changes can be induced by personal habits and lifestyle decisions (e.g., smoking, diet) as well. Equating genetic changes with illness, may encourage employers to view such employees as somehow disabled or unfit for work, making job discrimination a distinct possibility.

Genetic Screening

Option 5: Congress could amend section 504 of the Rehabilitation Act to prohibit discrimination in hiring against otherwise qualified applicants because their genetic screening results reveal a proclivity toward certain diseases in the future.

Amending section 504 in this manner would address several potential concerns. First, it tackles the problem of discrimination against job applicants, a topic left largely untouched by the OSH Act and NLRA protections. Second, it addresses what is perhaps the most likely area of abuse for the use of genetic screening. Third, it focuses on one of the possible uses of genetic screening, i.e., identification of applicants who are qualified but likely in the future to suffer from a disease that will require full use of sick leave or even early retirement. Finally, amending section 504 also permits Congress to address the use of genetic screening in the workplace to detect nonoccupationally related illnesses.

By focusing on this section, rather than with section 503, Congress could avoid the problem of directing employers to include those with genetic variants that do not otherwise qualify them as "handicapped" under their affirmative action programs.

The disadvantage, however, is the uncertainty associated with section 504's requirement that employers provide a reasonable accommodation for handicapped workers. It may be clear what accommodation is necessary to make a job accessible to one who is deaf or blind, and in turn to make a judgment whether that accommodation is reasonable to require of an employer. It may be more difficult, however, to judge what is necessary for someone with a currently asymptomatic genetic illness or susceptibility.

Option 6: Congress could direct the National Labor Relations Board to make preemployment genetic screening a mandatory subject of bargaining, in order to increase the possibilities for workers to protect themselves against what they and their representatives perceive as abuses of genetic screening.

Fitness-for-duty physicals and medical tests are already regarded as mandatory subjects of bargaining between unions and employers when applied to current workers. Thus, extending the concept to preemployment physicals and genetic screening

would not require markedly different concerns to be placed on the bargaining table, and would provide some protection for job applicants.

A disadvantage to this option, however, is that unions do not represent the majority of American workers, so this action would not protect all affected persons. Additionally, in light of interest in "two-tiered" systems of compensation, it is possible that unions and employers may trade protections for current workers from potentially discriminatory genetic screening tests for the privilege of screening job applicants more stringently.

ISSUE: Is the current Federal research agenda addressing genetic monitoring and screening adequately?

The current Federal research framework for addressing genetic monitoring and screening is composed of extramural and intramural programs sponsored by several agencies, including the Department of Energy (DOE), NIOSH, National Institutes of Health (NlH), National Center for Toxicological Research, Center for Environmental Health and Injury Control, and Agency for Toxic Substances and Disease Registry. OTA found that, in general, Federal research programs do not adequately address genetic monitoring or screening technologies for use in the workplace.

Option J: Take no action.

In the absence of congressional directives encouraging more research on the relationships between environmental exposures and health, information on gene-environment interactions will continue to be gathered piecemeal by the Federal agencies involved in this area. Some of the research funded by the National Center for Human Genome Research at NIH will contribute to the development of more valid and reliable tests. Research agendas of the National Cancer Institute and the National Institute of Environmental Health Sciences include studies relevant to this report. Work being conducted internationally is also contributing to knowledge in this area. Congress could decide that existing research capabilities will provide an adequate and appropriate level of information in this area and that no additional action needs to be taken. If Congress, however, decides that it is important to determine these relationships in order to provide for a safe and healthy workplace, and that the cost of occupational illness warrants more extensive examination, taking no action will result in incremental and disjointed progress in reaching these goals.

Option 2: Encourage the appropriate agencies to pursue studies that will provide a better understanding of the link between mutagenesis and carcinogenesis through larger, better controlled epidemiologic studies.

Over the years, an increasing number of health effects have been attributed to mutations caused by toxicants. The diseases most often associated with genotoxic substances are various forms of cancer. Mutational changes such as point mutations and chromosomal rearrangements have been associated with early stage tumor development. However, not all mutations cause disease. Because the relationship between mutation and health effect is often indirect and not well understood, more research needs to be conducted in this area. Epidemiologic studies in an occupational setting can address this problem.

Option 3: Direct the Secretary of the DOE to report on past and current research efforts directed by DOE toward identifying the genetic risks of radiation exposure.

Open-ended data collection by DOE, and **prob**lems of access to data, have stalled an open discussion of the real risks of radiation exposure to American workers in high- and low-level environments. DOE has recognized this problem and could be encouraged to share data with interested investigators.

Option 4: Ensure that the Food and Drug Administration (FDA) and the relevant offices of NIH properly evaluate new genetic monitoring and screening tests for reliability and validity.

There is some concern that the unique nature of many genetic diseases-which may present heterogeneity, reduced penetrance, and variable expressivity-pose significantly different challenges to diagnostic tests. Tests made available to employers or physicians need not only to be safe and effective, but to clearly explain the limitations in careful labeling so as to avoid misuse and misinterpretation. As more tests become available for both occupationally and nonoccupationally related diseases, issues of quality control must be addressed. NIH and FDA could cosponsor a Consensus Development Conference(s) on genetic monitoring and screening that could evaluate the accuracy, safety, labeling, and potential misuses and abuses of new tests.