Genetic Testing and Genetic Screening

PAT MILMOE MCCARRICK

In recent years there has been an enormous expansion in the knowledge that may be gleaned from the testing of an individual’s genetic material to predict present or future disability or disease either for oneself or one’s offspring. The Human Genome Project, which is currently mapping the entire human gene system, is identifying progressively more genetic sequencing information (see Scope Note 17, The Human Genome Project). Information obtained from genetic testing raises ethical and legal questions about its uses by society. The ethical dilemmas were foreseen two decades ago by bioethicists who asked whether questionable applications could stop “legitimate pursuits” (IV, Gaylin 1972) and whether genetic disease might come to be viewed as “transmissible” in the sense of being contagious (IV, Veatch 1974).

Not only has knowledge expanded, but the practice of genetic testing and screening has greatly increased as well. For example, in the case of testing for cystic fibrosis (CF), the U.S. Congress’ Office of Technology Assessment (OTA) estimates that instances of screening jumped from 9310 tests in 1991 to 63,000 tests in 1992 (I, OTA [Cystic Fibrosis] 1992). In addition, the OTA estimates that within the next few years, the six million women who become pregnant each year routinely will be informed of available CF carrier tests (I, OTA [Cystic Fibrosis] 1992).

Following the recent identification of a gene linked to breast cancer, Dr. Francis Sellers Collins, director of the National Center for Human Genome Research, said that “it is not inconceivable that every woman in America may want to be screened for this gene. The economic, ethical, and counseling issues will be very daunting.” Dr. Collins opines that in the near future physical examinations for 18-year-olds will include DNA testing for diseases with genetic components and that physicians, in the interests of preventive medicine, will make risk-based recommendations for a healthy life-style (IV, Breo 1993). The U.S. President’s Commission for the Study of Ethical Problems in Medicine and Biomedical and Behavioral Research predicted as early as 1983 that before the end of the century genetic screening and counseling would become major components of both public health and individual medical care (I, U.S. President’s Commission 1983).
The OTA defines genetic testing as "the use of specific assays to determine the genetic status of individuals already suspected to be at high risk for a particular inherited condition. The terms genetic test, genetic assay, and genetic analysis are used interchangeably to mean the actual laboratory examination of samples." In contrast, genetic screening usually uses the same assays employed for genetic testing but is distinguished from genetic testing by its target population (I, OTA [Genetic Monitoring] 1990). The National Academy of Sciences (NAS) defines screening as the systematic search of populations for persons with latent, early, or asymptomatic disease (III, NAS 1975). Some of the literature annotated for this Scope Note appears to use the terms "testing" and "screening" interchangeably.

Philip Boyle of the Hastings Center points out that the language used to describe genetic variation is important and asks what words should be used: "Defects, flaws, deleterious genes, disorders, or the more neutral conditions? Using words such as normal—and its corollary, abnormal—is likely to foster stigmatization and discrimination" (III, Genetic Grammar, Boyle 1992).

Areas of focus in genetic testing include: prenatal diagnosis, newborn screening, carrier screening, forensic screening, and susceptibility screening.

Prenatal diagnosis discerns whether a fetus is at risk for various identifiable genetic diseases or traits. Prenatal diagnosis is made using amniotic fluid, fetal cells, and fetal or maternal blood cells obtained during amniocentesis testing; alpha fetoprotein assays or chorionic villus sampling; or ultrasound tomography, which creates fetal images on a screen. Another method, known as fetoscopy, uses a camera on a needle inserted in the uterus to view the fetus. Since prenatal screening began in 1966 (I, U.S. President's Commission 1983), the number of metabolic defects and genetic disorders that can be diagnosed prenatally has expanded greatly. There is also discussion of requiring testing for parents who are participating in an in vitro fertilization program and are at genetic risk. Preimplantation testing of embryos might ensure that only embryos free of genetic disease or problem traits would be placed in the uterus (IV, Robertson 1992).

Newborn screening involves the analysis of blood or tissue samples taken in early infancy in order to detect genetic diseases for which early intervention can avert serious health problems or death. Newborn screening first came into use in the early 1960s with the ability to test newborns for a rare metabolic disease, phenylketonuria (PKU), which causes mental retardation and can be prevented by following a special diet (III, Genetics and the Law, Capron 1990). Two other examples of newborn screening, in place since the 1970s, are the testing of African-American infants for sickle cell anemia and Ashkenazic Jews for Tay-Sachs disease (IV, Reilly 1991).

Carrier screening identifies individuals with a gene or a chromosome abnormality that may cause problems either for offspring or the person screened. The testing of blood or tissue samples can indicate the existence of a particular genetic trait, changes in chromosomes, or changes in DNA that are associated with inherited diseases in asymptomatic individuals (I, OTA [Genetic Monitoring] 1990). Groups tested include persons at risk or a cross-section of the general public for occurrence statistics. Examples of carrier screening include the previously mentioned tests for
sickle cell anemia and for Tay-Sachs disease. In the last few years, screening tests have also been developed for cystic fibrosis, Duchenne muscular dystrophy, hemophilia, Huntington's disease, and neurofibromatosis (II, March of Dimes Birth Defects Foundation 1992; IV, Breo 1993). Recently it also has become possible to identify certain cancer prone individuals through genetic testing (IV, Li et al. 1992).

Forensic testing, which is the newest area to use information obtained from genetic testing, seeks to discover a genetic linkage between suspects and evidence discovered in criminal investigations. Test results have been presented as proof of innocence or guilt in court cases, and jury verdicts have been based on this type of genetic evidence. Critics note that forensic laboratories often test just once, unlike research laboratories, which test many times, and that mistakes can be made (IV, Hoeffel 1990). Concern is expressed, too, about the confidentiality of DNA profiles obtained from criminal investigations and stored in national police databanks (IV, Bereano 1990). Debate now centers on standards and quality control, but it is accepted that the technologies accurately detect genetic differences between humans and are "new, powerful tools to clear the innocent and convict the guilty." Since DNA is unique, many people are reluctant to see such information become part of any national database, which might include information not only about identity but also about proclivity toward disease or behavior (I, OTA [Genetic Witness] 1990).

Finally, susceptibility screening is used to identify workers who may be susceptible to toxic substances that are found in their workplace and may cause future disabilities. In 1986, Morton Hunt wrote in the New York Times Magazine that 390,000 workers become disabled by occupational illness each year; he thinks these illnesses are precipitated by genetic hypersusceptibility since co-workers are unaffected (IV, Hunt 1986).

In an early classic work, the National Academy of Sciences says screening can be used for medical intervention and research; for reproductive information; for enumeration, monitoring, and surveillance; and for registries of genetic disease and disability (III, NAS 1975). Many factors affect the use of any routine screening: customs of care (including both professional guidelines and possible malpractice); education of the public about the results and limitations of genetic testing; availability, training, and education of personnel to perform testing; financing of such screening (particularly third-party payor responsibilities); stigmatization and discrimination issues; quality assurance of laboratories and DNA test kits; and costs and cost effectiveness (I, OTA [Cystic Fibrosis] 1992).

The Committee of Ministers of the Council of Europe thinks that the public generally recognizes the benefits and the potential usefulness of genetic testing and screening for individuals, for families, and for the population as a whole, but it says that there is an accompanying anxiety that genetic testing and screening arouses. Its recommendations to allay any future unease include: informing the public in advance; educating professionals to provide quality services (genetic tests would only be carried out by physicians); offering appropriate, non-directive, counseling; providing equality of access; respecting the self-determination of those tested; making testing or screening non-compulsory; and denying insurers the right to
require testing or to seek the results of previous tests (I, Council of Europe 1992).

The Danish Council of Ethics views genetic information as different from other private information since it reveals knowledge not only about an individual, but also the individual's relatives, and because analyses will provide comprehensive information about both individuals and population groups. The Council says that screening provides information useful either to the individual or to public health officials, but this information is not concerned with treatment. From a public health point of view, testing may prevent costly treatment of a disease, protect third parties, and give the person the option of treatment. However, from the individual's point of view, there may be ambivalence about the possibility of a relative's potential disease (I, Danish Council of Ethics 1993).

Not everyone thinks that the growing field of genetic testing and screening is beneficial. The potential problems raised both by those who favor testing and screening and those who oppose it are similar, but one faction thinks that regulatory or legislative solutions to the problems can be found while concerned opponents find the knowledge itself less valuable and the problems unsolvable. Opponents of widespread genetic testing and screening regard the acceptance of eugenic theories and scientists' inability to control outcomes of their genetic research as dangerous. They foresee a need to outlaw technologies that threaten privacy or civil rights and a need to protect against genetic discrimination. "We need to engage in active debates about the practical consequences of genetic forecasts for our self-image, our health, our work lives, our social relationships, and our privacy" (III, Hubbard and Wald 1993). Disability advocates and feminists have criticized genetic screening because they think it fosters intolerance for less than perfect people (IV, Kristol 1993).

Another possible negative effect is the pressure that might be placed on individuals, as a result of cost-benefit analysis, to test or to be tested. Individuals might thereby be forced to know their genetic predispositions, to tell others, or to act to save society long-term costs resulting in a "new eugenics based, not on undesirable characteristics, but rather on cost-saving" (I, Knoppers 1991). However, Lowe (IV, 1991) points out that genetic testing will not create more illness than presently exists, and it could lead to a reduction in costs due to early treatment. Lippman (V, 1992) suggests that control over genetics would create an elite who could control the general populace, particularly if mandatory testing or intervention were viewed as a community good. Other potential adverse effects of such screening include the development of prejudice against those tested and found at risk and the feeling of tested persons that they are predetermined victims of fate or are being branded as "abnormal" (I, Danish Council on Ethics 1993).

Fletcher and Wertz (III, Genetics and the Law, 1989), who have surveyed geneticists throughout the world, say that "the dangers of isolation, loss of insurance, educational, and job opportunities for persons diagnosed with incurable and costly disorders known from early childhood are real to many who are concerned about potential clinical uses and abuses of the 'new genetics.'"

Guidelines and suggestions for ways to avoid negative effects that may arise from genetic testing are prominent in the literature. Significant proposals include the duty
of authorities to help those persons identified to be at risk, the need for respect in personal areas, the right of autonomy, and the realization that a newly diagnosed genetic problem may create difficulties in an individual's relationships with others. In any genetic screening, guidelines should be established governing its aim, limitations, scope, and ethical aspects, as well as the storage and registration of data or material, the need for followup (including social consequences), and the risk of side effects (I, Danish Council on Ethics 1993). Genetic screening should always be voluntary, not mandatory, according to 99 percent of those surveyed by the OTA with reference to cystic fibrosis screening (I, OTA [Cystic Fibrosis] 1992).

Issues of confidentiality loom large in discussions of genetic testing and screening. According to the Privacy Commission of Canada, genetic privacy has two dimensions: protection from the intrusions of others and protection from one's own secrets. It concludes that privacy is an explicit constitutional right that includes respect for genetic privacy and is protected by legislation. Consequently, employers, in general, should be prohibited from collecting genetic information; services and benefits should not be denied on the basis of genetic testing; and information should be used only to inform a person's own decisions (I, Privacy Commission of Canada 1992).

The President's Commission, in a 1983 study, concluded that genetic information "should not be given to unrelated third parties, such as insurers or employers, without the explicit and informed consent of the person screened or a surrogate for that person." The Commission recommended that information stored in computers should be coded and that compulsory genetic screening cannot be justified to create a health gene pool or to reduce health costs (I, U.S. President's Commission 1983). More recently, the NIH/DOE Working Group on the Ethical, Legal and Social Implications of Human Genome Research recommended that health insurers should consider a moratorium on the use of genetic tests in underwriting (I, NIH/DOE 1993).

In the area of data protection and professional secrecy, genetic information for health care, the diagnosis or prevention of disease, and for research should be stored separately from other personal records. In addition, those handling the information should be bound by professional rules of confidentiality and legislative rules, and any unexpected findings should be given only to the person tested (I, Council of Europe 1992).

The literature on genetic discrimination suggests several areas of sensitivity: (1) the workplace, where employers may choose to test job applicants, or those already employed, for susceptibility to toxic substances or for genetic variations that could lead to future disabilities, thereby raising health or workmen's compensation costs; (2) the insurers (either life or health insurance companies) who might use genetic information or tests as criteria for denying coverage or require reproductive testing to be done for cost containment purposes; and (3) law enforcement officials, who may test and/or use information without informed consent (II, American Medical Association 1991). Thomas H. Murray thinks that "genetic testing in the workplace was a putative public health measure in its old form and now is used as a means of saving money or promoting health." He opines that access to genetic testing
involves considerations of justice since genetic testing competes with other scarce resources and it may emphasize racial and ethnic differences (II, AAAS/ABA 1992).

In nationwide surveys, 80% of the public indicates that it expects genetic technology to be beneficial, 71% thought that it would pose risks to them and their family, and 62% thought the benefits outweighed the risks (I, OTA [Genetic Tests] 1990). A 1992 Louis Harris poll found that 68% of the persons questioned knew little or nothing about genetic testing, but 79% would undergo testing prior to having children to learn whether a child might inherit a fatal genetic disease (II, March of Dimes 1992). Strict regulations are favored by 75% of those polled (II, March of Dimes 1992).

Regulations that offer some protection in the areas of discrimination and confidentiality were listed in the OTA's Genetic Monitoring and Screening in the Workplace and include:

- The National Sickle Cell Anemia Control Act [P.L. 92-294], amended in 1976 to the National Sickle Cell Anemia, Cooley's Anemia, Tay-Sachs, and Genetic Diseases Act [P.L. 94-278]. The laws increased research, training, testing, counseling, and education in the area of screening for sickle cell anemia and other genetic diseases.

As early as the 1970s, the National Academy of Sciences looked at legal principles and raised questions about the extent of disclosure of test results to the person screened, the extent of disclosure to others without the consent of the person screened, the constitutional barriers to mandatory screening by states, and the constitutional difficulties encountered if screening is done by racial or ethnic group (III, NAS 1975). In a 1992 report, OTA offers six areas for possible action by Congress: genetics education for the public; genetics training and education of health care professionals; discrimination (access to health care coverage); laboratory and other regulation; means of automating diagnostics; and facilitating use of genetic assays in clinical practice (I, OTA [Cystic Fibrosis] 1992).

In an article aimed at family physicians, Howard Stein (V, 1992) writes of the physician-patient relationship and reminds his fellow physicians that: "Genetic knowledge does not occur in a social vacuum. The scientific account is neither the only story, nor the entire story. Decisions to know or not to know, to have children or not to have children, to label as diseased or not, are part of wider life histories, language, and group fantasies." Legal challenges, government regulation, extensive education, and collective bargaining will all be part of the ongoing process needed to solve the complex dilemmas foreseen to be part of future widespread genetic testing and genetic screening (III, Genetic Grammar, Draper 1992).

The report examines how to “benefit from the potential of genetic technology without undermining our autonomy” and “spawning another nightmare in our surveillance society.” Part I provides a description of the scientific fundamentals of genetic testing with present applications; Part II discusses privacy principles relevant to both the public and private sectors; Part III examines the Privacy Act’s regulation of genetic testing; Part IV considers regulation of private sector genetic testing; and Part V draws conclusions. The appendix contains a summary of positions on genetic testing and privacy taken by other countries and by international organizations. Twenty-two recommendations are presented.


Recommendations for public acceptance and understanding of genetic testing and screening are presented along with guidelines concerning public and professional education and the assertion that research projects must conform with medical ethics standards.


The publication contains three separate reports: protection of sensitive personal information; genetic screening and its ethical aspects; and finally, a section on genetic testing, which includes a copy of a bill that would forbid genetic testing in the workplace and in insurance underwriting. The National Board of Health outlines 14 screening guidelines.


Divided into six chapters, the first two provide an overview of genetics, the genome, and the gene pool. The next two discuss human dignity and genetics from an international viewpoint and from that of the Canadian Charter of Rights and Freedoms. The final chapters present material on genetic determinism and naturalism, genetic discrimination (including workplace, insurance, and reproductive testing), genetic justice, and ethical considerations and principles. The document contains a glossary as well as tables of Canadian and American cases, statutes, treaties, and international agreements.

Containing recent statistics on genetic testing for cystic fibrosis, OTA reports that present testing misses 5 to 15 percent of the carriers. A survey of commercial insurers indicated that 90 percent of respondents would accept an applicant with a family history of cystic fibrosis at standard rates. OTA concludes that the value of the CF carrier test is the information it provides but that it cannot estimate what it means to individuals to know. The test sensitivity is in the 85 to 90 percent range; almost half of those surveyed thought that 95 percent sensitivity should be required before widespread CF carrier screening is offered.

The report describes issues involved in genetic monitoring and screening in the workplace, examines the technologies, analyzes the legal aspects, assesses the ethical issues, discusses the role of genetic counseling, and evaluates current and future uses. The report surveyed 1500 companies and unions and evaluates the current and future use of genetic monitoring and screening in the workplace. The main concern of employees and applicants is protection of their autonomy and privacy, keeping all information confidential, especially if could be used to deprive them of a job, health insurance, or other benefits.

OTA surveyed commercial insurers (Blue Cross/Blue Shield plans and health maintenance organizations) to assess their views and practices toward reimbursement for genetic tests and their policies in using test results in underwriting.

The paper reports the results of a nationwide survey of public knowledge and opinion about issues in science and genetics, particularly risk, gene therapy, and the future of such technology.

OTA found that a Florida criminal conviction based on DNA typing greatly raised interest in using such tests and, at the same time, raised concerns about infringement of civil liberties. Linkage of information in public and private sources is seen as equivalent to creating a national database, raising issues of
informational privacy. Questions of data security, quality, and reliability are discussed.


The task force recommends that health status information should not be used to deny health care coverage or services to anyone. They think that genetic services should be comparable to other health services and would include testing, counseling, and treatment. Health insurers should “consider a moratorium on the use of genetic tests in underwriting.”


The President's Commission cautions about the “subtle interplay of social norms and individual choices required as genetic screening and counseling become increasingly important.” The Commission's basic conclusion is that genetic screening is a valuable service when established with concrete goals and procedural guidelines founded on ethical and legal principles.

### II. ORGANIZATIONS


The conference pointed out that while the human genome project will lead to more reliable, less expensive testing for an increasing number of genetic characteristics, it will also raise profound issues of ethics, law, and social policy. The book is an overview of the conference discussions along with three papers commissioned for the conference: The Scientific Basis of the ‘Genetic Revolution’: A Selective Review, by Paul R. Billings; The Human Genome Project and Genetic Testing: Ethical Implications, by Thomas H. Murray; and Legal Issues in Genetic Testing, by John A. Robertson.


The Council points out that employers, insurers, and law enforcement agencies all have uses for genetic information and techniques. It concludes that gener-
ally it is inappropriate to exclude workers because of risk, but a limited testing might be done: (1) if a disease were so rapidly serious and irreversible that monitoring could not prevent harm; (2) if data demonstrate that abnormality results in many susceptible persons; or (3) if the cost of lowering the toxic substance to protect the susceptible is too high. Testing must not done without the informed consent of the employee or applicant for employment.


The Society did not reach consensus that widespread screening should be carried out for cystic fibrosis. However, members agreed: that carrier testing for CF should be offered to couples when a close relative has CF; that several pilot programs should be conducted to gather data; that laboratories conducting tests should have centralized quality control; that large-scale population screening should begin when tests detect a larger proportion of CF carriers and provide more information; and finally, that routine CF carrier testing of pregnant women and other individuals is not a standard of care.


The Foundation commissioned Louis Harris and Associates to poll the public to learn more about its knowledge of genetic testing. It found that significant minorities of Americans are opposed to taking genetic tests and express a measure of fear, but that the majority of those polled would undergo genetic testing. Interestingly, 41 percent said that they or their family have some form of genetic or inherited health problem.

III. BOOKS/SPECIAL ISSUES


Andrews provides a full discussion of the legal ramifications of medical genetics. She includes sections on genetic research, particularly embryo and fetal research; informed consent and duties to disclose; wrongful birth and wrongful life; confidentiality; and mandatory screening.


Lawyer Andrews discusses access to genetic services and confidentiality of information, pointing out that legal liability will undoubtedly upgrade physicians' knowledge and use of genetic technologies.


Saying that there are three levels of issues in the Human Genome Project, Annas describes the first level as genetic screening and counseling on the individual/family level with negligence
in offering or performing them properly resulting in lawsuits. The second level involves societal issues such as population-based genetic screening, resource allocation and commercialization, and eugenics. He thinks the third level involves issues of determinism, reductionism, normalcy, and the meaning of health and disease. Annas suggests ways to regulate genetic technologies and their possible future uses.


Capron discusses four kinds of genetic screening: newborn screening, carrier screening, prenatal testing, and susceptibility screening.


Focusing on genetic screening for cystic fibrosis, Robertson says there is concern whether and how such screening programs should be implemented. He discusses preimplantation screening of embryos and the ethical issue of embryo discard.


In a survey of 677 international geneticists, the authors asked respondents to rank the significance of ten ethical issues. The study provides guidelines for ethical problems that may arise in medical genetics and for avoiding abuse of genetic knowledge by employers, insurers, or the health care system.


Physician Elsas poses a number of ethical and moral questions that genetic technologies raise in the physician-patient relationship. He includes potential legal liabilities and some ethical-legal questions in genetic responsibility, and he suggests some principles of genetic screening.


Boyle, Philip J. Introduction. P. S1.


Nolan provides information about the ways that genetic information may change the nature of pregnancy and suggests the need for national standards and criteria for normal prenatal care.


The author questions the social and ethical implications of genetic diagnosis of embryos in IVF, asking who will monitor the technique and whose needs are met? She thinks research has not been accompanied by an openness about applications.

Rothman, Barbara Katz. Not All That Glitters Is Gold. Pp. S11-S15. Imagining a level of quality control in procreation that could make baby farms possible, Rothman warns of the dangers of preimplan-
tation genetic diagnosis and the high physical, psychological, and social costs to the prospective mother.


Draper considers discrimination in the workplace and in insurance coverage, using cases as examples. She predicts that new policies about genetic information will only follow complex legal challenges, government regulation, education, and collective bargaining.


A young alcoholic asks his physician to determine whether his drinking is genetic. The authors comment on the case and its consequences for employment and health insurance.


The authors hold that a new genetic determinism serves a conservative social policy that blames ill health on individuals rather than on environment or social conditions. They stress the threat to privacy and civil liberty that can result from genetic prediction.


This early classic in the field of genetic screening provides an ongoing framework to study the prospects, history, and development of principles, legislation, and program guidelines applicable to genetic testing aims, methodology, and education. Ethical aspects are presented from the view of a "perfect" screener, who would have all relevant facts to provide both error-free testing and effective counseling; possess a strong sense of the thoughts and emotions of those screened; be as free as possible from self-interest and inappropriate emotionalism; and apply principles consistently.


Wertz and Fletcher report and discuss "a cross-cultural study of the approaches of medical geneticists to ethical problems in genetic counseling, prenatal diagnosis and screening." The authors surveyed 682 genetics professionals from 19 countries to obtain the widest possible views, and they note that all contacted "stressed the need to protect the privacy of tested people from institutional third parties, especially insurance companies and employers." Fourteen clinical cases about five different ethical situations were circulated to obtain views about full disclosure of sensitive information, access of family members and others to genetic information, prenatal diagnosis, fair access to genetic services, workplace disease susceptibility, access of information in the workplace, screening for cystic fibrosis, and presymptomatic testing for Huntington's disease. Fletcher reviews the survey in a final chapter on ethics and human genetics.
Nations represented in the survey include: Australia, Brazil, Canada, Denmark, Federal Republic of Germany, France, German Democratic Republic, Greece, Hungary, India, Israel, Italy, Japan, Norway, Sweden, Switzerland, Turkey, United Kingdom, and United States.

IV. JOURNAL ARTICLES/CHAPTERS


Saying that one in 25 white persons in Britain and the United States carries the cystic fibrosis gene, the authors recommend screening partners in sequence, screening the second only if the first is positive.


Bereano thinks that civil liberty problems are inherent in DNA identification systems, and he offers principles such as taking samples from suspects only after obtaining a search warrant or court order and storing only fingerprints (not samples). A copy of the state of Washington’s American Civil Liberties Union Board of Directors’ statement on DNA extraction and analysis is included.


The authors say that genetic discrimination is based on a variation from the “normal” human genotype and that such discrimination already exists in the health and life insurance industries, citing denial of services or entitlements to persons found to be asymptomatic or to those who may never be impaired. They fear a new “social underclass” based on genetic discrimination and recommend changes in social attitudes, legal protection, and the health care system to prevent genetic discrimination from growing.


The author poses various possible dilemmas including a case of a preemployment physical that would show that the applicant has a susceptibility to cancer, asking “does he have a right to protect such information or to choose his own risk level?” Screening indicates genetic intervention could be made, according to Bohrer, and he asks when the state can intrude in the welfare of a patient or when costs will control the distribution of genetic benefits.


In a theme issue on molecular genetics, the author reports that the field of gene hunting is moving very rapidly as evidenced by the discovery of the genes that cause cystic fibrosis (1989), neurofibromatosis (1990), and Huntington’s disease (1993), as well as the ongoing research in the laboratory
of Francis Sellers Collins, director of the National Center for Human Genome Research, for the causes of inherited breast cancer, the cause of a form of leukemia, and the cause of a form of sudden cardiac death from arrhythmia.


Comparing genetic research to a shopping spree, Brom says that employers and insurers will use the vehicle of genetic testing to seek out the perfect pair of genes in order to avoid adverse selection and costly health insurance. She urges legislation that would limit the use of genetic information by these groups, asserting that otherwise large numbers of people will be defined as unemployable or uninsurable and their health costs will be borne by society.


The author agrees with certain hospital protocols that recommend that testing for Huntington's disease not be done if a patient is psychologically frail. Calling this view a principle of minimal paternalism, he provides ethical decision-making theory relevant to suicidal persons, alcoholics, schizophrenic or manic depressive persons, the extremely retarded, and autistic persons who might be excluded from genetic testing (see also IV, MacKay 1991; Quaid 1991).

Fletcher, John C.; Berg, Kare; and Tranoy, Knut Erik. Ethical Aspects of Medical Genetics: A Proposal for Guidelines in Genetic Counseling, Prenatal Diagnosis and Screening. *Clinical Genetics* 27 (2): 199-205, 1985.

The authors propose guidelines for different aspects of genetic screening: mandatory screening (except for newborns with treatable disorders) is objectionable; test values should be demonstrated; children should not be screened although adolescent screening may confer benefits in reproductive choices and planning; screening information should be given to third parties only with the consent of the screened; and genetic information contained in public and private databases should be protected by state-of-the-art security.


Frieden asks if genetic knowledge will mean that only a healthy few will have health insurance, and she provides anecdotal information on several of the 40 cases of discrimination collected by geneticist Paul Billings. She says that most cases of insurance discrimination result from access to tests taken rather than companies requesting such tests since mass screening is costly. Problems include the accuracy of tests, interpretation of results, and how and by whom the information will be used.


In this early article, Gaylin urges wise application of any new technology and deplores paternalism in keeping information from those tested.

Gevers, Sjef. Use of Genetic Data, Employment and Insurance: An Interna-
Medical testing in relation to employment or underwriting of insurance has been practiced, the author says, and he thinks that the new genetic technology will aggravate problems since medical data and genetic data can be seen as similar. Discovery of genetic traits showing serious disorders could lead to social stigma and potential discrimination according to Gevers.


Glasser raises questions about genetic information obtained in criminal investigations, asking whether it will be destroyed, or if stored, where it will be kept and who might have access to it. In the workplace, if genetic susceptibility is determined, he thinks employers may remove the worker, not the hazardous substance. He urges codification of enforceable legal limits before there is large scale collection of genetic information.


Harper recounts the long history of the genetic study of Huntington’s disease, including compulsory sterilization laws and the eugenic views practiced in Nazi Germany. He thinks that serious abuse may evolve from computerized genetic registers or inappropriate testing and urges preparation to avoid future dangers.


The author finds that little use has been made of genetic testing for insurance purposes but notes that three groups are particularly concerned about this possibility: insurance companies, those tested or likely to be tested, and medical geneticists, each of whom view the issue differently. Harper recommends that population genetic screening be avoided if there are insurance implications and that companies should have the right to ask about relevant genetic tests in large or unusual life insurance applications, but not for “normal” policies or for health insurance.


Reporting on a meeting at the National Center for Human Genome Research at the National Institutes of Health, the article discusses the meaning of abnormality, genetic discrimination, differential treatment, and eugenics. Meeting attendee Patricia King says that genetic determinism is already used to justify bigotry; when genetic differences are collected by race or ethnicity, further discrimination will result. Other questions raised at the meeting were: If quick and reliable tests are available, will all physicians have to test or be liable for omission and who will explain the computerized readouts given to patients?


In a reprint of a speech given to the insurance industry, Hoar says that ge-
Netic technology is both expensive and very popular. Since insurance companies use the general population as a whole to determine risk estimates and actuarial tables, high risk individuals are buried in the whole. Testing identifies high risk and may be good for insurance, which is a business. He thinks that “it is a bag of worms...and that we are now on what you might call a genetic tightrope.”


In a discussion of the use of DNA material in court cases, Hoeffel says that juries often weigh statistical evidence differently and come to different conclusions depending upon presentation. She thinks that the government could easily move from databanks with criminal DNA profiles to larger databanks with DNA profiles of all citizens. She can imagine that DNA profiles could be accessible not only to law officials but to insurance companies, employers, schools, adoption agencies, and other organizations who would “need to know” “in the public interest.”


Saying it would be a major boon if each person could be searched or screened for vulnerability to environmental toxicity, Hunt writes that 390,000 workers become disabled by occupational illness each year and that 100,000 die each year. He thinks that this is due to genetic hypersusceptibility, since co-workers are unaffected, and that what is true in the workplace is true everywhere. Testing conflicts with our notions of equality and fairness, according to Hunt, who outlines early industry testing from 1970 to 1982. He discusses various moral dilemmas: (1) equal opportunity versus health protection (quoting Judith Areen, who says genetic testing can create de facto discrimination); (2) equal opportunity versus free enterprise; (3) fairness to the handicapped versus “the greatest happiness of the greatest number”; (4) individual freedom versus social control; (5) knowledge versus privacy; and (6) paternalism versus autonomy. The author recommends working out a set of compromise regulations after consulting all factions.


Justifications offered in support of programs for cystic fibrosis screening include prenatal diagnosis to make decisions about a CF pregnancy; neonatal screening for early diagnosis and treatment; and carrier screening to make marriage and reproductive decisions. The authors urge that screening goals and the target population be well defined; that laboratory quality control be stringent, with limits of results clearly delineated; that authorities protect the confidentiality of the information; that procedures to protect individual and family privacy be established in advance; that participation be voluntary; that genetic counseling be offered and that educational programs are in place; and that long-term outcomes be monitored and evaluated.

Kristol, Elizabeth. Picture Perfect: The

Kristol writes that genetic prenatal testing has grown because it places physicians in the best possible legal position in the event of a birth defect and because the public health sector has an interest in reducing morbidity and mortality rates. She says that those in policy positions will be forced to judge what is too costly for society and what kind of life is worth living.


The authors suggest ten guidelines for screening newborns, saying that such screening is a medical act that should lead to universally available medical intervention. Other principles they find important include: informing parents of the significance of the findings, having follow-up procedures in place, respecting confidentiality, and possibly using blood specimens for anonymous research.


Predictive testing for the germ-line mutations should be guided by the ethical principles of respect for autonomy, beneficence, confidentiality, and justice according to the authors. All persons should be given “current relevant information on the test to make an informed voluntary decision.” “The right to decide to undergo testing rests solely with the individual concerned,” and information obtained should not be given to others without the consent of the person or the guardian. They say that testing should be provided regardless of finances; that participants should be able to withdraw at any time, but should be encouraged to follow-up after the test result is known; and that counseling and support services should be in place before testing begins. Compliance with these guidelines should minimize “psychological, social, economic, and other harm that might result from predictive p53 testing.” Nonetheless, the authors conclude that the benefit of testing cannot be assumed.


Lippman expresses concern that shaping definitions of illness and health, normality and abnormality, have not been adequately analyzed and that doing so will reinforce inequities in the distribution of health care and will legitimize a new area for social control.


Even prior to the expected boom in genetic testing, the author says that insurers discriminate against those genetically predisposed to illness. He outlines health insurance practices in the United States, including risk pools and self-insuring. The loss of those who learn that they are a good risk and who wish low premium insurance could cause industry problems, but he thinks an equilibrium would be reached rapidly.

MacKay discusses autonomy with a view to the ultimate good of the person including the need to “do no harm,” use due care, and consider the risk-benefit ratio along with the benefit-detriment equation. (See also IV, DeGrazia 1991; Quaid 1991.)


Markel says that stigmatization and ostracism of those who are found to have “undesirable” traits after genetic screening could increase. He compares screening to quarantine, saying that healthy society separated itself from the “ill,” and addresses two episodes when genetics were applied to American social policy: the early 20th century eugenics movement and the 1970s screening programs for sickle cell anemia.


The authors studied three groups: the first was screened and found to be carriers of the Tay-Sachs gene; the second was screened and did not have the Tay-Sachs gene; and the third was a control group, which was not screened. All viewed their present and past health in the same manner, but the carrier group was less optimistic about the future. The authors urge study of people’s screening experience before undertaking any mass genetic screening programs.


Saying that few laws explicitly regulate the treatment of genetic information, McEwen and Reilly indicate that the nation’s legislatures have many such acts under consideration, reflecting “a growing societal awareness that the uncontrolled dissemination and use of genetic data entails significant risks.” Their broad 50-state survey provides information about current law and includes: statutes that call for informed consent and that protect the confidentiality of various types of genetic information; law that prohibits both insurers and employers from requiring or administering a genetic test; law that regulates or prohibits information reaching insurance companies; and law that prohibits genetic discrimination by life and disability insurers. The article includes specific citations to state laws.


The authors describe possible discriminatory uses of genetic tests by employers and insurance companies, and they note that instances have been reported. Federal and state statutes and court decisions that can be used for protection against such discrimination are cited. The authors predict that genetic discrimination will become more prevalent as genetic testing becomes more common. They think that current legislation adequately covers genetic discrimi-
nation in employment but that it does not prohibit discrimination by insurance companies.

Post, Stephen G. Huntington's Disease: Prenatal Screening for Late Onset Disease. *Journal of Medical Ethics* 18 (2) 75-78, June 1992.

Post discusses the selective abortion of fetuses on the basis of prenatal screening for late-onset genetic diseases. He concludes that it is difficult to justify ethically although it remains a matter of personal choice.


The author says that 200 individuals were tested for Huntington's Disease in the United States between 1986 and the time she wrote the article with no disastrous outcomes (e.g. suicide). The majority of those at risk have chosen not to be tested. Aside from the high cost of testing, she sees the main barrier as limitations in treatment of the disease. (See also IV, DeGrazia 1991; MacKay 1991.)


Reilly says the first experience in carrier testing in the United States was in the early 1970s with poorly conceived state laws governing testing programs for the sickle cell trait. A federal law corrected the problems and offered funding to state programs that respected privacy, provided counseling, and were voluntary. A better 1970s community screening program was devised to identify the Tay-Sachs disease gene. Today about 25,000 Ashkenazic Jews in the United States are tested each year, and the number of children born with this disease is about one-tenth that of the pretesting time. Reilly recommends pilot testing and counseling projects be put in place for cystic fibrosis.


Robertson discusses technology whereby embryos from in vitro fertilization could be screened genetically before placement in the uterus if a couple were at risk for serious genetic disease. Only embryos without disease would be implanted in the uterus. Robertson notes that this preimplantation genetic screening may be viewed as extensive manipulation of embryos with a potential for eugenic selection of offspring and a positive engineering of offspring traits. He discusses the moral status of the embryo, embryo discard, embryo research, active alteration of embryos, and legal aspects of preimplantation screening.


Rothstein states that genetic tests for cancer susceptibility raise difficult legal, ethical, and policy issues affecting medical treatment, public health policy, insurance and employment screening, occupational and environmental regulation, commercial applications, and liability issues. He writes that current laws were not designed to regulate genetic testing and that no national consensus on the ethical and health policy implications of genetic testing has been developed. Observing that genetic screen-
ing for cancer susceptibility may introduce mass screening, fetal screening, interest from life and health insurance corporations, and application to the entire field of reproduction or universal premarital testing, Rothstein recommends funding for research on genetic testing; consideration of whether it is appropriate to require, permit, or prohibit tests for cancer susceptibility; and safeguards necessary to ensure privacy of such information, particularly to employers and insurers.


The paper is based on a review of 119 articles and provides developmental background on genetic testing as well as an outline of the ethical problems identified (including objectives, selection of candidates, and the nature of the techniques involved) and solutions adopted in preclinical protocols.


In this early work, Veatch examines value orientations in genetics, looking at the emerging fields of genetic counseling, mass screening, and gene therapy from the view of different ethical theories.


The authors' list of privacy and disclosure problems from genetic knowledge includes: six cases arising from genetic tests that disclosed false paternity to an unsuspecting husband; disclosure of a person's genetic make-up to a spouse; disclosure, against a patient's wishes, to relatives at risk; ambiguous test results; disclosure of unexpected nonmedical information such as fetal sex; and disclosure of genetic information to institutions, e.g. employers and insurers. The authors write about an ethics of responsibility to care for persons, and they develop such an approach in their case studies.


Physician Whittaker says that as genetic screening increases, there will not be enough geneticists to meet demand and that primary care physicians will be responsible for the delivery and evaluation of these tests as a part of medical practice. She discusses applications, confidentiality, and legal and malpractice issues, concluding that it is important for physicians to appraise and understand the therapeutic relevance and implications of the tests and to set policy along with scientists, ethicists, legislators, and the lay public.

V. ADDITIONAL READINGS:


Blank, Robert H. Politics and Genetic Engineering, Politics and the Life Sci-


SCOPE NOTE SERIES

The SCOPE NOTE Series is intended to present a current overview of issues and viewpoints related to specific topics in biomedical ethics. It is not designed as a comprehensive review, but rather offers immediate reference to facts, opinion, and legal precedents (if applicable) for scholars, journalists, medical and legal practitioners, students, and interested laypersons.

All sources cited in SCOPE NOTES are included in the collection of the National Reference Center for Bioethics Literature, and may be obtained through its document delivery service (subject to copyright law). Updates of topics covered in the SCOPE NOTE Series may be obtained by searching the BIOETHICSLINE database (accessed through the National Library of Medicine’s MEDLARS system); or BIOETHICSLINE Plus, Silver Platter’s CD-ROM version of the database; or by calling the National Reference Center for Bioethics Literature.

As noted in the list below, some of the SCOPE NOTES have appeared in the Kennedy Institute of Ethics Journal (KIEJ); each is published separately as a reprint and is available for $5.00
each prepaid from: National Reference Center for Bioethics Literature, Kennedy Institute of Ethics, Georgetown University, Washington, DC 20057-1065 or telephone 1-800-MED-ETHX (toll-free) or 1-202-687-6738. (Add $3 each for airmail outside North America.) Series editor: Doris Mueller Goldstein. The following SCOPE NOTES are presently available:


No. 4. Diagnosis Related Groups (DRGs) and the Prospective Payment System: Forecasting Social Implications. June 1984. 11 p.


