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SCOPE

NOTE

22

Genetic Testing and Genetic Screening

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First published in September 1993, Scope Note 22 is an annotated bibliography with links to electronic texts where possible. It is updated on a periodic basis.

Background Essay

The completion of the mapping of the human genome (see [Scope Note 17: The Human Genome Project](#)) shifts the spotlight of ethical inquiry from general questions about genetic research to specific issues with such topics as genetic screening. Ethical dilemmas with genetic testing and screening were foreseen over three decades ago by bioethicists who asked whether questionable applications could stop "legitimate pursuits" (1, [Gaylin 1972](#)) and whether genetic disease might come to be viewed as "transmissible" in the sense of being contagious (1, [Veatch 1974](#)).

The use of genetic tests has increased greatly as knowledge has expanded. One early example was screening for cystic fibrosis (CF). In [Cystic Fibrosis and DNA Tests: Implications for Carrier Screening](#), the U.S. Congress' Office of Technology Assessment (OTA) reported that instances of screening jumped from 9,310 tests in 1991 to 63,000 tests in 1992 (6, [OTA 1992](#)). OTA's prediction that CF carrier tests would be offered routinely to the six million women who become pregnant each year (6, [OTA 1992](#)) was realized when the National Institute of Health's [consensus statement](#) on the topic recommended CF testing for adults with a positive family history of the disease, for partners of people with CF, for couples currently planning a pregnancy, and for couples seeking prenatal care (6, [NIH 1997](#)). General [guidelines](#) for genetic testing were issued by the NIH Task Force on Genetic Testing in 1997 (1, [NIH Task Force](#)

1997). In response to a further recommendation of the Task Force, the [Secretary's Advisory Committee on Genetic Testing \(SACGT\)](#) was established in June, 1998, by the Department of Health and Human Services to provide oversight of all aspects of genetic testing. SACGT's [report](#) on oversight issues was published in June, 2000 (1, [SACGT 2000](#)).

In 1983, the [report](#) of the U.S. President's Commission for the Study of Ethical Problems in Medicine and Biomedical and Behavioral Research predicted that before the end of the century genetic screening and counseling would become major components of both public health and individual medical care (1, [U.S. President's Commission 1983](#)). Following the identification of a gene linked to breast cancer, BRCA1, 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." (6, [Breo 1993](#)). While genetic testing for BRCA mutations has been available commercially since 1996, the results of an [evaluation](#) done by the Centers for Disease Control and Prevention (CDC) indicated that population-based screening "...is not recommended because of the complexity of test interpretation and limited data on clinical validity and utility." (8, [CDC 2004](#)) In 2005, the United States Preventive Services Task Force concurred with the CDC and [recommended](#) that only women with a family history of breast and ovarian cancers be referred for BRCA mutation testing. (8, United States Preventive Services Task Force) Educational materials about gene tests and international directories of clinics and laboratories providing genetic testing services can be found on the Web site [GeneTests](#) sponsored by the National Institutes of Health, as well as on [MedlinePlus: Genetic Testing](#) located on the U.S. National Library of Medicine's Web site.

As a greater proportion of the U.S. population lives beyond 85 years of age, interest in genetic testing for end-of-life conditions such as Alzheimer Disease (AD) continues to grow. Stephen G. Post observes that the "too-hopeful" general public has assigned a degree of scientific certainty to the as yet preliminary genetic findings for AD, and that teaching critical thinking skills about genetic testing to the general public is of the highest priority [8, [Post and Whitehouse 1998](#)].

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 (5, [OTA 1990](#)). The National Academy of Sciences (NAS) defines screening as the systematic search of populations for persons with latent, early, or asymptomatic disease (1, [NAS 1975](#)). Some of the literature annotated for this Scope Note appears to use the terms "testing" and "screening" interchangeably.

Philip Boyle 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" (1, [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 (1, [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 (7, [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 (1, [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 (1, [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 (5, [OTA 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 (1, March of Dimes Birth Defects Foundation 1992; 6, [Breo 1993](#)). Recently it also has become possible to identify certain cancer prone individuals through genetic testing (1, [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 (4, [Hoeffel 1990](#)). Concern is expressed, too, about the confidentiality of DNA profiles obtained from criminal investigations and stored in national police databanks (5, [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 (4, [OTA 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 (5, [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 (1, [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 (6, [OTA 1992](#)).

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" (1, [Hubbard and Wald 1993](#)). Disability advocates and feminists have criticized genetic screening because they think it fosters intolerance for less than perfect people (7, [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" (1, [Knoppers 1991](#)). Now that British insurers have government approval to use the results of screening for Huntington's disease to assess insurance premiums, consumer groups say that individuals will be reluctant to have

such tests and risk denial of coverage (3, [Dickson 2000](#)). On the other hand, [Lowe \(3, 1991\)](#) 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 \(6, 1992\)](#) 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" (1, [Danish Council on Ethics 1993](#)).

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 (5, [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 (1, [AAAS/ABA 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](#) (5, [OTA, 1990](#)). The National Human Genome Research Institute maintains a [database](#) of current federal and state regulations focusing on the protection of genetic information.

In an article aimed at family physicians, Howard Stein (1, [Stein 1992](#)) writes of the physician-patient relationship and reminds his fellow physicians that "[g]enetic 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 that result from widespread genetic testing and genetic screening (1, [Genetic Grammar, Draper 1992](#)).

1. Overview

American Association for the Advancement of Science. *The Genome, Ethics and the Law: Issues in Genetic Testing*. A Report of a Conference on the Ethical and Legal Implications of Genetic Testing sponsored by the AAAS-ABA National Conference of Lawyers and Scientists and the AAAS Committee on Scientific Freedom and Responsibility, Coolfont Conference Center, Berkeley Springs, West Virginia, June 14-16, 1991. Washington: American Association for the Advancement of Science, 1992. 124 p.

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.

American Society of Human Genetics. **Statement on Cystic Fibrosis Screening**. *American Journal of Human Genetics* 46(2): 393, February 1990.

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.

Andrews, Lori B. ***Future Perfect: Confronting Decisions About Genetics***. New York, Columbia University Press, 2001. 264 p.

Noting the wide impact of genetic testing, Andrews discusses patient self concept, reproduction decisions, confidentiality, discriminations in health insurance and employment, the need for competent counseling, and the legal, ethical and social regulations she deems important to help solve dilemmas arising from the tests and their ensuing ramifications. She describes three models, pointing out both advantages and problems: a medical model with physician gatekeepers, a public health model that could provide education as well some testing for those unable to afford tests, and a fundamental rights model that could give greater weight to individuals decisions about the use of health care services. The book has 85 pages of footnotes.

Andrews, Lori B. ***Medical Genetics: A Legal Frontier***. Chicago: American Bar Foundation, 1987. 284 p.

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.

Annas, George J. **Mapping the Human Genome and the Meaning of Monster Mythology**. *Emory Law Journal* 39(3): 629-664, Summer 1990.

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.

Asch, David A.; Patton, James P.; Hershey, John C.; and Mennuti, Michael T. **Reporting the Results of Cystic Fibrosis Carrier Screening**. *American Journal of Obstetrics and Gynecology* 168(1, Part 1): 1-6, January 1993.

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.

Bekker, Hilary; Denniss, Gill; Modell, Michael; Bobrow, Martin; and Marteau, Theresa. **The Impact of Population Based Screening for Carriers of Cystic Fibrosis**. *Journal of Medical Genetics* 31(5): 364-368, May 1994.

The study looks at patients aged 18 to 45 in an inner London general practice: 637 women and 329 men who agreed to be tested for the cystic fibrosis gene. Positive results caused anxiety, but the authors say it almost disappeared within three months. They conclude that population screening's greatest problem is one of "false reassurance" rather than anxiety.

Bissell, Michael G. **Ethics and Professionalism: The Case of Genetic Testing**. *Clinical Laboratory Management Review* 8(5): 501-506, September-October 1994.

Discussing prenatal and carrier screening, Bissell cites the clinical laboratory code of ethics: professionalism, respect for persons, economic accountability, scientific integrity, and social responsibility, stressing that genetic testing will be a major part of laboratory work in the next century.

Boyle, Philip. **Introduction to Genetic Grammar: 'Health,' 'Illness,' and the Human Genome Project**. *Hastings Center Report* 22(4): S1, July-August 1992.

Boyle introduces the essays in this Special Supplement in the *Hastings Center Report*. Authors Kathleen Nolan, Andrea Bonnicksen, and Barbara Katz Rothman (section 7 Table of Contents); Elaine Draper and Paul Billings (section 5) are included.

Capron, Alexander Morgan. **Which Ills to Bear: Reevaluating the 'Threat' of Modern Genetics**. Emory Law Journal 39(3): 665-696, Summer 1990.

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

Clarke, Angus; Fielding, David; Kerzin-Storarr, Lauren; et al. **The Genetic Testing of Children**. Report of the Working Party of the Clinical Genetics Society (UK). Journal of Medical Genetics 31(10): 785-797, October 1994.

The working party recommends predictive genetic testing of children when the condition occurs in childhood or when treatment can be offered. For adult onset disease, it advises against testing when the child is healthy and no medical intervention is useful although the family could discuss it and children could choose testing when they are "autonomous adults." The party urges further research when "there is insufficient evidence to know whether a diagnosis in childhood is helpful in the medical management of the possibly (not yet) affected child." Saying that testing children is for carrier status is more complex, the working party would make a presumption against testing except in certain specific circumstances. Adoptive situations are also discussed.

Council of Europe. Working Party on Genetic Testing and Screening. **Draft Recommendation on Genetic Testing and Screening for Health Care Purposes and Draft Explanatory Memorandum**. Adopted by the Ad Hoc Committee of Experts on Bioethics (CAHBI), Strasbourg, September 2-3, 1991. For the attention of the Committee of Ministers (accepted by the Committee of Ministers, October 2, 1992.) 18 p.

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.

Danish Council of Ethics. **Ethics and Mapping of the Human Genome**. Copenhagen: Danish Council of Ethics, 1993. 86 p.

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.

Drlica, Karl A. **Double-Edged Sword: the Promises and Risks of the Genetic Revolution**. Reading, MA: Addison-Wesley Publishing Company, 1994. 242 p.

Decrying complex jargon, the author discusses genetic concepts in layperson's terms. Saying that each of us will be faced with genetic problems in our lifetime, Drlica discusses prenatal diagnosis, predisposition to health problems, and genetic tracking in an effort to provide a "framework for evaluating new applications of genetics."

European Commission. **[Ethical, Legal and Social Aspects of Genetic Testing: Research, Development and Clinical Applications](#)**. Eryl McNally (chair) and Anne Carbon-Thomsen (coordinator). Brussels, 2004. 127 p.

Based on [25 recommendations](#) made by an expert panel convened by the European Commission, this report covers such topics as "genetic exceptionalism", counseling for genetic conditions, biobanks and confidentiality, and population screening. A conference for citizens and stakeholders was held in Brussels on May 6th -7th 2004 to discuss the implementation of the recommendations contained in the report.

European Community Huntington's Disease Collaborative Study Group. **Ethical and Social Issues in Presymptomatic Testing for Huntington's Disease: A European Community Collaborative Study**. Journal of Medical Genetics 30(12): 1028-1035, December 1993.

Studying four major genetic testing centers in different European Community countries, the study group identified the issues as problems of inappropriate referral, problems involving relatives, and problems relating to

disclosure of results. They stress the importance of expert counselling and of close liaison between clinical, counselling, and laboratory staff.

Fibison, Wendy J. **The Nursing Perspective: Genetic Screening and Testing.** *Journal of School Nursing* 9(4): 16-21, 23-24, December 1993.

Fibison urges school nurses to provide leadership in preparing for genetic decisions that young people and their families will have to make regarding possible future genetic screening and testing. She says nurses need to be aware of tests, counseling services and the "ethical, legal, and social implications."

Fletcher, John C., and Wertz, Dorothy C. **Ethics, Law, and Medical Genetics: After the Human Genome Is Mapped.** *Emory Law Journal* 39(3): 747-809, Summer 1990.

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.

Frankel, Mark S.; and Teich, Albert H., eds. **The Genetic Frontier: Ethics, Law, and Policy.** Washington: American Association for the Advancement of Science (AAAS), 1994. 240 p.

Two of the four sections in this report of a conference sponsored by the AAAS-American Bar Association National Conference of Lawyers and Scientists and the AAAS Committee on Scientific Freedom and Responsibility are concerned with genetic testing. Part II includes privacy and confidentiality issues discussed in three areas by Laurence Tancredi, Alan F. Westin, and Madison Powers. Part IV reports on genetic testing and determination of property rights: one by Gilbert S. Omenn on the scope of patent protection; three papers touching on intellectual property and genetic testing by Kate H. Murashige, Thomas J. White, Joan Overhauser; and Ted Peters, intellectual property and human dignity.

The Future of Medicine: How Genetic Engineering Will Change Us In the Next Century. *Time* 153(1): 11 January 1999 [Special Issue].

This issue devoted to a broad view of the entire field of genetics offers several specific essays about the ramifications of genetic testing/screening: Good Eggs, Bad Eggs: The Growing Power of Prenatal Genetic Tests Is Raising Thorny New Questions About Ethics, Fairness and Privacy by Frederic Golden, David Bjerklie, and Alice Park, pp. 56-59; Playing the Odds: Health Insurers Want to Know What's in Your DNA by Christopher Hallowell, p. 59; and The DNA Detectives by Jeffrey Kluger, pp. 62-63.

Gaylin, Willard. Genetic Screening: **The Ethics of Knowing.** *New England Journal of Medicine* 286(25): 1361-1362, 22 June 1972.

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

Great Britain. Parliament. House of Commons. Committee on Science and Technology. **Human Genetics: The Science and Its Consequences.** Volume I. Reports and Minutes of Proceedings. 145 p. **Human Genetics.** Volume II. Memoranda received up to January 31, 1995. 197 p. **Human Genetics.** Volume III. Memoranda received after January 31, 1995. 58 p.

The report describes the science of human genetics, the genome project and genetic research in the United Kingdom, medical applications of genetics, genetic science and industry, and human rights including discrimination, privacy, employment and insurance, ethical, legal and social issues, and international regulations of human genetics. There is a nine page summary of conclusions and recommendations (Vol. I, pp. xci-xcix); including the establishment of a Human Genetics Commission with statutory powers "to deal with screening and related matters."

Grody, Wayne W., and Pyeritz, Reed E. **Report Card on Molecular Genetic Testing: Room for Improvement?** *JAMA: Journal of the American Medical Association* 281(9): 845-847, 3 March 1999.

The authors emphasize that the scope of molecular genetic testing is increasing so rapidly that professional medical societies must work to ensure high quality in the process and physicians must prepare to be involved both before and after testing "to a high degree." They think that molecular testing will be the predominant way that physicians of all specialties will diagnose and manage patients well into the 21st century.

Hepburn, Elizabeth R. **Genetic Testing and Early Diagnosis and Intervention: Boon or Burden?** *Journal of Medical Ethics* 22(2): 105-110, April 1996.

Hepburn urges that the assessment of the value of the information to those involved should be considered along with the severity of the disability and the accuracy of the data when persons receive genetic testing diagnoses. Both treatment success and prognosis are important indices of its value. She offers three approaches: public health screening when tests are accurate and treatment is reasonable and simple; comprehensive educational programs targeted at specific groups prior to childbearing when there will be transmission of a defective gene resulting in serious and untreatable birth disabilities; and finally, to offer honest information and sensitive assistance in decision making to those whose genetic factors show a predisposition to a disease for which there is no curative treatment.

Holtzman, Neil A. **The UK's Policy on Genetic Testing Services Supplied Direct to the Public-- Two Spheres and Two Tiers.** *Community Genetics* 1998 1(1): 49-52.

In the Code of Practice on Human Genetic Testing Services Supplied Direct to the Public prepared by the United Kingdom's Advisory Committee on Genetic Testing, the public can obtain testing for inherited recessive disorders direct from suppliers or commercial laboratories. For other types of genetic testing a physician must be involved. Thus some tests are available to those who can pay for them; others must obtain testing through the National Health Service which at present covers the testing of relatives for cystic fibrosis, but not the general public. The short, seven point code is available on p. 48.

Hubbard, Ruth, and Wald, Elijah. ***Exploding the Gene Myth: How Genetic Information Is Produced and Manipulated by Scientists, Physicians, Employers, Insurance Companies, Educators, and Law Enforcers.*** Boston: Beacon Press, 1993. 206 p.

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.

Institute of Medicine. ***Assessing Genetic Risks: Implications for Health and Social Policy.*** Washington, DC: National Academy Press, 1994. 338 p.

The report contains recommendations about "acquiring and using genetic information in health care in a manner that respects the autonomy of individuals." The committee notes the legal ramifications of rights to genetic information and that this report "should not be interpreted as creating a set of legal guidelines." The committee recommends a "significant increase in genetics education " for both health personnel and the public, and a "centralized oversight to ensure that new genetic tests are accurate and effective, that they are performed and interpreted with close to 'zero-error' tolerance, and that the results of genetic testing are not used to discriminate against individuals in employment or health insurance."

Kapp, Marshall B. **Ethical and Legal Implications of Advances in Genetic Testing Technology.** *In Legal Medicine 1994.* Cyril H. Wecht, ed. Salem, NH: Butterworth Legal Publishers, 1994, pp. 305-319.

Kapp says that although genetic testing may have current shortcomings, the testing will surely improve in terms of reliability and that the ethical and legal implications should be faced soon. He urges health professionals and individuals considering testing to realize that the ethical dilemmas faced by families and businesses must be

taken into consideration, both legally and ethically. He discusses duty to be tested, right/duty to receive results, duty to share results versus right of confidentiality, employers and insurers, duty to accept interventions, who pays for tests and treatment, and autonomy versus obligation.

Kimyai-Asadi, Arash; and Terry, Peter B. **Ethical Considerations in Pulmonary Genetic Testing and Gene Therapy**. *American Journal of Respiratory and Critical Care Medicine* 155(1): 3-8, January 1997.

The authors predict that pulmonologists will be involved in complex decision making as genetic testing and treatment develops, requiring "a greater understanding of the medical and social implication of genetic disorders." Outlining scientific process as descriptive (when characteristics are recognized), informational (when genes are identified and linked to disease), and treatment phase, they think there can be a chronologic gap between phases. Gene identification is in its "infancy," raising ethical questions of informed consent, confidentiality, and selective abortion based on genetic information. Opining that the next concern for physicians will be gene therapy, they present guidelines for genetic testing and gene therapy.

Knoppers, Bartha Maria. **Human Dignity and Genetic Heritage**. Study Paper prepared for the Law Reform Commission of Canada in the Protection of Life Series. Ottawa: Law Reform Commission of Canada, 1991. 93 p. (Also in French, 103 p.)

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.

Lehrman, Sally. **Genetic Testing 'Needs More Checks.'** *Nature* 378(9): 120-121, 9 November 1995.

Lehrman reports that scientists attending the annual meeting of the American Society of Human Geneticists, called for "a system to evaluate the accuracy and interpretations of tests, as well as turn-around times." Saying that "academic and commercial laboratories are rushing to offer genetic testing with a minimum of external review" she cites anecdotal cases and several studies.

Li, Frederick P.; Garber, Judy E.; Friend, Stephen H.; et al. **Recommendations on Predictive Testing for Germ Line p53 Mutations Among Cancer-Prone Individuals**. *Journal of the National Cancer Institute* 84(15): 1156-1160, 5 August 1992.

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 benefits of testing cannot be assumed.

Lippman, A. **Led (Astray) by Genetic Maps: The Cartography of the Human Genome and Health Care**. *Social Science and Medicine* 35(12): 1469-77, December 1992.

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.

March of Dimes Birth Defects Foundation. **Genetic Testing and Gene Therapy: National Survey Findings**. White

Plains, NY: March of Dimes, September 1992. 18 p.

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.

Marteau, Theresa M.; van Duijn, Monica; and Ellis, Ian. **Effects of Genetic Screening on Perceptions of Health: A Pilot Study**. *Journal of Medical Genetics* 29(1): 24-26, January 1992.

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.

McCrary, S. Van; Allen, Bill; Moseley, Ray; Crandall, Lee A.; et al. **Ethical and Practical Implications of the Human Genome Initiative for Family Medicine**. *Archives of Family Medicine* 2(11): 1158-1163, November 1993.

The authors, writing for a special Human Genome Insurance Project conducted at the University of Florida College of Medicine, say that predictive genetic testing will "change significantly the routine practice of family medicine." Issues of informed consent, confidentiality, and physician record keeping are discussed and illustrative cases concerned with the "potential impact of these emerging technologies" are included.

McGovern, Margaret M., Benach, Marta O., Wallenstein, Sylvan, et al. **Quality Assurance in Molecular Genetic Testing Laboratories**. *JAMA: Journal of the American Medical Association* 281(9): 835-840, 3 March 1999.

In a survey of 245 molecular genetic testing laboratories, the authors indicate that many were operating sub-optimally and in need of improvement in personnel qualification and practice standards. Particular attention focused on the need to guard patient rights to confidentiality and informed consent.

Murray, Thomas H. **Assessing Genetic Technologies: Two Ethical Issues**. *International Journal of Technology Assessment in Health Care* 10(4): 573-582, Fall 1994.

The author discusses ten factors that characterize the social context of contemporary genetics, considering "two presumptions that usually are unquestioned: first, that more choice is always better; second, that what can be improved should be improved." Saying that genetic screening and testing can be an ambiguous good as prenatal genetic testing grows longer, he says the tests offer "no way to distinguish between significant disease and parental whim...Genetic technologies increasingly will challenge the troubled distinction between therapy and enhancement."

Murray, Thomas H. Murray; Rothstein, Mark A.; and Murray, Robert F., Jr., eds. **The Human Genome Project and the Future of Health Care**. Bloomington, IN: Indiana University Press, 1996. 248 p.

The work offers ideas about practical health care applications of the Human Genome Project's findings. Various authors discuss the physician patient relationship and the necessity for primary care physicians to become genetic experts, prenatal diagnosis, issues of employment and health insurance, the use of genetic testing results to allocate access to care, and the way policy will be set according to genetic data.

National Academy of Sciences and National Research Council. **Genetic Screening: Programs, Principles and Research**. Washington: National Academy of Sciences, 1975. 388 p.

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.

National Institutes of Health (United States) [NIH]. Secretary's Advisory Committee on Genetic Testing (SACGT). [Enhancing the Oversight of Genetic Tests: Recommendations of the SACGT](#). Bethesda, MD: National Institutes of Health, 2000. 42 p.

National Institutes of Health (United States) [NIH] - U.S. Department of Energy. Working Group on Ethical, Legal, and Social Implications of Human Genome Research. [ELSI] Task Force on Genetic Testing (Neil A. Holtzman, chair) [Promoting Safe and Effective Genetic Testing in the United States: Principles and Recommendations](#). September 1997. [<http://www.genome.gov/10001733>]

The Task Force reviewed both the operational and ethical issues raised by the commercial availability of genetic testing. Their final report includes discussions of genetic testing validity, of informed consent for genetic counseling, and of the relationship between evidence-based medicine and genetic screening. It contains the Task Force's recommendations on quality assurance measures for laboratories performing genetic tests and on the need for a national body with the authority to review genetic testing practices.

Nobles, Kimberley. **Birthright or Life Sentence: Controlling the Threat of Genetic Testing**. *Southern California Law Review* 65(4): 2081-2128, 1992.

Saying that confidentiality, consent, counseling, reliability, and verification must be in place in any genetic testing program, lawyer Nobles states that existing law offers some protection, but that "lack of regulation in testing has paved the way for abuse and discrimination." "Testing programs and informational access must be strictly monitored to ensure that an individual's constitutional rights are safeguarded."

Nuffield Council on Bioethics. **Genetic Screening: Ethical Issues**. London: Nuffield Council on Bioethics (28 Bedford Sq., London WC1B 3EG), December 1993. 115 p.

In his preface to the report, chairman Sir Patrick Nairne says that genetic research differs from many medical advances because of the speed of its development, its effect on individuals, families and the general society, and also the "fear it arouses that it may be interfering with the basis of life itself." He highlights features included in the report: the difficulty assessing individual health risks exposed by screening, affecting both insurance and employment; the complexity of maintaining professional patient confidentiality; the demands on resources and quality; and need to "safeguard against potential eugenic abuse." The comprehensive report discusses genetic counseling, informed consent, disclosure to individuals and family members, confidentiality, employment, insurance, public policy and ways to implement programs in place in the United Kingdom.

Parker, Lisa S.; and Majeske, Rachel Ankeny. **Standards of Care and Ethical Concerns in Genetic Testing and Screening**. *Clinics in Obstetrics and Gynecology* 39(4): 873-884, December 1996.

The authors think that standards of care are not just medical or scientific in genetic testing and screening, differing in that they are substantially "ethical questions that involve balancing risks and possible benefits to patients in light of their goals and values and in light of complex social practices like insurance." They discuss cancer risk, breast cancer screening, cystic fibrosis, and issues of informed consent. They conclude that traditional ethical concerns (psychological sequelae, discrimination risks, false information, and false security) are part of any developing standard.

Phoenix, David D.; Lybrook, Sherrill M.; Trottier, Ralph W.; Hodgin, Faye Cobb; and Crandall, Lee A. **Sickle Cell Screening Policies as Portent: How Will the Human Genome Project Affect Public Sector Genetic Services?** *Journal of the National Medical Association* 87(11): 807-812, November 1995.

The authors looked at sickle cell screening as "an example of a technology that was introduced in a manner that raised poignant issues." They urge "discussion and development of a national consensus on appropriate content

and just delivery of public sector genetic services." Cultural sensitivity is cited as of considerable importance in delivering genetic service to avoid stigmatization based on race or religion. They think that each county and state should review newborn screening regulations and legislation to ensure that they are accurate and current and to not imply discriminatory treatment.

Public Priorities for Genetic Services. Special Supplement. Hastings Center Report 25(3): S1-S24, May-June 1995.

The supplement looks at advances in genetic information and testing, analyzing the "multitude of unresolved ethical issues." Two articles discuss screening, urging "caution on those responsible for setting priorities" and two other articles discuss the legal and the commercial aspects of various genetic services.

Quaid, Kimberly A. **Predictive Testing for HD: Maximizing Patient Autonomy.** *Journal of Clinical Ethics* 2(4): 238-239, Winter 1991.

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, Philip R. **Advantages of Genetic Testing Outweigh Arguments Against Widespread Screening.** *Scientist* 5(2): 9, 11, 21 January 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.

Singapore. Bioethics Advisory Committee. Genetic Testing and Genetic Research. Singapore: The Bioethics Advisory Committee, November 25, 2005. 394 p. Available online: <http://www.bioethics-singapore.org/resources/reports4.html>

Based on an extensive public consultation process involving the general public, health care professionals, and religious leaders, Singapore's Bioethics Advisory Committee (BAC) issued recommendations covering both clinical genetic testing and genetic research. Special attention is given to preimplantation testing and counseling of vulnerable populations.

Stein, Howard F. **The Human Genome as Metaphor.** *Journal of Family Practice* 35(3): 256-258, September 1992.

Sommerville, Ann, and English, Veronica. **Genetic Privacy: Orthodoxy or Oxymoron?** *Journal of Medical Ethics* 1999 April; 25(2): 144-150.

While noting that all patient decisions should take into account responsibilities to family members and to the community, the authors discuss the ways in which genetic testing highlights issues of autonomy and patient choice. Sommerville and English conclude by asking the question "Can genetic information be exclusively owned?"

Stone, David H.; and Stewart Susie. **Screening and the New Genetics: A Public Health Perspective on the Ethical Debate.** *Journal of Public Health Medicine* 18(1): 3-5, March 1996.

Noting that the two most frequently cited objectives of genetic screening for recessive carrier state genes are to reduce prevalence and to inform individuals or couples at risk, the authors say that this "represents a paradigm shift in the philosophy of screening in that no preventive principle is involved." Information is regarded as worthwhile regardless of the outcome, and the authors draw "attention to the danger that a combination of

technical capability, professional zeal, and consumer demand will override currently acceptable screening principles. In this event, future efforts to subject screening programs to rational evaluation could be undermined."

Takala, Tuija. **The Right to Genetic Ignorance Confirmed.** *Bioethics* 1999 July; 13(3-4): 288-293.

The author rebuts the argument that "...genetic information about oneself is also information about one's relatives," and asserts that individuals have the right to refuse to know the results genetic testing even when the request for testing is made for a pedigree study.

Terrenoire, Gwen. **Huntington's Disease and the Ethics of Genetic Prediction.** *Journal of Medical Ethics* 18(2): 79-85, June 1992.

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.

U.S. Congress. Office of Technology Assessment (OTA). **New Developments In Biotechnology: Background Paper--Public Perceptions of Biotechnology.** Washington: Government Printing Office, 1987. (OTA-BR-BA-45, S/N 052-003-01068-2.) 127 p.

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.

U.S. President's Commission for the Study of Ethical Problems in Medicine and Biomedical and Behavioral Research. **Screening and Counseling for Genetic Conditions: A Report on the Ethical, Social, and Legal Implications of Genetic Screening, Counseling, and Education Programs.** Washington: Government Printing Office, 1983. 122 p. [<http://bioethics.georgetown.edu/documents/pcemr/geneticscreening.pdf>]

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.

Veatch, Robert M. **Ethical Issues in Genetics.** Chapter 7 in *Progress in Medical Genetics*, Volume X, ed. Arthur G. Steinberg and Alexander G. Bearn, pp. 223-264. New York: Grune & Stratton, 1974.

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.

Wertz, Dorothy C.; and Fletcher, John C. **Proposed: An International Code of Ethics for Medical Genetics.** *Clinical Genetics* 44(1): 37-43, July 1993.

Presenting arguments for and against adopting a medical genetics code, along with the eight ethical problems they see as evident in medical genetics, Wertz and Fletcher discuss reproductive choices, confidentiality problems within families, protection of privacy from insurers, employers, and government agencies, disclosure dilemmas, controversial indications for prenatal diagnosis, voluntary or mandatory screening, and counseling.

When is Genetic Screening Permissible? Editorial. *Nature* 377(6547): 273, 28 September 1995.

Saying that the "World Medical Association is planning to fashion a statement on the ethical principles of genetic screening from a draft that is altogether over-cautious", the Nature editorial questions as false any assumption that patients should not receive "potentially disquieting information" and suggests that more awareness will lead to a more "rational attitude towards the inevitability of death of which modern societies (and health-care systems) are in great need."

Wilfond, Benjamin S.; and Nolan, Kathleen. **National Policy Development for the Clinical Application of Genetic Diagnostic Technologies: Lessons from Cystic Fibrosis.** *JAMA Journal of the American Medical Association* 270(24): 2948-2954, 22/29 December 1993.

Commenting on the rapid advances in genetic knowledge, the authors write that it is possible that every test that is feasible will be routinely given to promote commercial interests, allay legal liability, or satisfy patient's demands. They urge an evidence based model developed from the evaluation of research, utilizing both professional and the public understanding of issues to set a standard of care.

Zimmern, R. L. **Genetic Testing: A Conceptual Exploration.** *Journal of Medical Ethics* 1999 April; 25(2): 151-156.

Positing that there is nothing essentially different about genetic information that requires special regulation, Zimmern acknowledges that "...fear and mistrust of DNA technology by the public requires that society act to regulate the technology itself."

2. Privacy/Discrimination

American Society of Human Genetics. Social Issues Subcommittee on Familial Disclosure. **Professional Disclosure of Familial Genetic Information.** *American Journal of Human Genetics* 62(2): 474-483, February 1998.

The ASHG statement outlines points to consider: the general rule of confidentiality, exceptional circumstances that permit disclosure, and the ethical duty to inform patients about familial implications. Background discussion includes: ethical frameworks for disclosure of otherwise confidential information, the duty to warn under law, and international trends and positions.

Andrews, Lori B. **Introduction to Genetics and the Law.** Randolph W. Thrower Symposium, Emory University School of Law, April 4, 1990. *Emory Law Journal* 39(3): 619-628, Summer 1990.

Attorney 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.

Annas, George J. **Genetic Prophecy and Genetic Privacy--Can We Prevent the Dream from Becoming a Nightmare?** [Editorial]. *American Journal of Public Health* 85(9): 1196-1197, September 1995.

The author holds that the new genetics "raises virtually every major health care policy questions as well as unique legal and ethical problems." Annas thinks that genetic information is "uniquely private and personal" for three reasons "it can predict an individual's likely medical future; it divulges personal information about one's parents, siblings, and children; and it has a history of being used to stigmatize and victimize individuals." Urging federal legislation to protect privacy, he offers a draft genetic privacy act "the core of which prohibits individuals from analyzing DNA samples unless they have verified that written authorization for the analysis has been given by the individual or his or her representative."

Boetzkes, Elisabeth. **Genetic Knowledge and Third-Party Interests.** *Cambridge Quarterly of Healthcare Ethics* 8(4): 386-392, Summer 1999.

Protection from the interests of insurers or prospective employers are discussed from the point of view of the ethical basis of the professional obligation to confidentiality as well as the right to privacy of those tested. However, the author supports family members, saying that "first-party resistance to disclosure may be overcome by weighty third-party claims."

Burnside, John W. **Ethical Quandaries in Genetic Testing.** *Texas Medicine* 93(2): 46-49, February 1997.

Burnside calls the increased availability of genetic information new science, but says that the old dilemmas of disclosure, value of information, who owns the information, unexpected information problems, special problems in testing children, and who should receive information still remain. He concludes that the information belongs to the patient, information has value beyond medical decision making, that confidentiality should only be breached with "patient waiver or with a very compelling threat of danger to others," and that unexpected information should be given to the patient. Patients need to know the "implications of testing, but they do not need to be told whether or not to test." Burnside says that he will not "participate in testing for nonmedical genetic markers."

Canada. Privacy Commission of Canada. *Genetic Testing and Privacy*. Ottawa: Minister of Supply and Services, 1992. 111 p. (Also in French, 111 p.).

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 rational organizations. Twenty-two recommendations are presented.

Couser, G. Thomas. **Genome and Genre**. In his *Vulnerable Subjects: Ethics and Life Writing*. London and Ithaca, NY: Cornell University Press, 2004, pp. 165 - 197.

Referring to Alice Wexler's account of her mother's diagnosis and death from Huntington's disease in [Mapping Fate](#), Couser recommends personal narratives as ... "a reality check on the 'genohype' that has surrounded the Human Genome Project." Noting that our "...knowledge of genetics outstrips [our] understanding of it," the author posits that Wexler's book counters the stigma associated with genetic disease "...by demystifying her mother's illness and recuperating her as a subject with some agency, rather than a passive victim of a rogue gene."

Epstein, Richard A. **The Legal Regulation of Genetic Discrimination: Old Responses to New Technology**. *Boston University Law Review* 74(1): 1-23, January 1994.

Defining genetic discrimination as "discrimination directed against an individual or family based solely on an apparent or perceived genetic variation from the 'normal' human genotype", Epstein holds that genetic discrimination "raises problems no different from those associated with any other sort of misfortune, and calls for no different response." He thinks that full disclosure to insurer or employer is appropriate and that "today's dominant mode of analysis does not pay so much as a passing nod to the lessons of contracting economics or the logic of public choice."

Goldworth, Amnon. **Informed Consent in the Genetic Age**. *Cambridge Quarterly of Healthcare Ethics* 8(3): 393-400, Summer 1999.

Amnon stresses the importance of autonomy and the need to give informed consent by anyone who undergoes genetic testing. He thinks that "it should be difficult to trump autonomy by consequential claims," and wonders "how much individual self-governance the citizens of a democratic society will be willing to forgo in order to achieve a collective good."

Rhodes, Rosamond. **Genetic Links, Family Ties, and Social Bonds: Rights and Responsibilities in the Face of Genetic Knowledge**. *Journal of Medicine and Philosophy* 23(1): 10-30, February 1998.

Rhodes looks at individuals responsibilities to one another in light of genetic testing, using three cases to illustrate her views. She concludes that no one has a moral right to genetic ignorance, and that moral responsibility "depends on a variety of factors including blood ties, social relationships, the history of interaction, and particular features of the situation and the individuals involved."

Robertson, John A. **Procreative Liberty and Human Genetics**. *Emory Law Review* 39(3): 97-719, Summer 1990.

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.

Harper, Peter S. **Huntington Disease and the Abuse of Genetics**. *American Journal of Human Genetics* 50(3): 460-464, March 1992.

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.

Henig, Robin Marantz. **Pitfalls of Genetic Screening: New Techniques Raise Difficult Ethical Questions**. *Washington Post Health* 7(5): 14, 29 January 1991.

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?

Jonsen, Albert R. **Genetic Testing, Individual Rights, and the Common Good**. In *Duties to Others*, Volume 4, Theology and Medicine Series. Courtney S. Campbell and B. Andrew Lustig, eds. Dordrecht, The Netherlands, Kluwer Academic Publishers, 1994, pp. 279-291.

Jonsen says that modern genetics point up the genetic commonalities that make all persons more alike than different and goes on to discuss confidentiality versus the obligation to reveal genetic information, and privacy and personhood versus the family's ownership of such information. Calling four features relevant he describes the clinical transaction that generates information, the severity of the condition, the burden the patient is likely to experience, and the implications from having this diagnosis.

Kerem, Eitan, and Lynch, Abbyann. **Screening for Cystic Fibrosis: Ethical and Social Issues**. *American Review of Respiratory Disease* 143(3): 457-461, March 1991.

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.

Markel, Howard. **The Stigma of Disease: Implications of Genetic Screening**. *American Journal of Medicine* 93(2): 209-215, August 1992.

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.

McEwen, Jean E, and Reilly, Philip R. **State Legislative Efforts to Regulate Use and Potential Misuse of Genetic Information**. *American Journal of Human Genetics* 51: 637-647, 1992.

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.

Natowicz, Marvin R.; Alper, Jane K.; and Alper, Joseph S. **Genetic Discrimination and the Law**. *American Journal of Human Genetics* 50(3): 465-475, March 1992.

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 discrimination in employment but that it does not prohibit discrimination by insurance companies.

Wachbroit, Robert. **The Question Not Asked: The Challenge of Pleiotropic Genetic Tests**. *Kennedy Institute of Ethics Journal* 8(2): 131-144, June 1998.

Moving on from single-gene disorders, the author is concerned with complex genetic mutations where testing may indicate that more than one disease could develop. He asks whether patients being tested for one condition should be informed if another possible condition is found, and concludes that all information should be disclosed.

Wertz, Dorothy C., and Fletcher, John C. **Privacy and Disclosure in Medical Genetics Examined in an Ethics of Care**. *Bioethics* 5(3): 212-232, July 1991.

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.

Wiesenthal, David L.; and Wiener, Neil I. **Privacy and the Human Genome Project**. *Ethics and Behavior* 6(3): 189-202, 1996.

Calling the reduction in cost of genetic testing an unintended consequence of the Human Genome Project, the authors express concern that "privacy as we know it is dead." They think that the "combination of scientific breakthroughs, free market capitalism, and the desire for new ways to deal with medical and social problems will accelerate the use of genetic data." They urge development of an "ethics of privacy" since others may know more about an individual than that person knows of herself or himself.

Wolf, Susan M. **Beyond "Genetic Discrimination": Toward the Broader Harm of Geneticism**. *Journal of Law, Medicine & Ethics* 23(4): 345-353, Winter 1995.

Commenting on gender or race genetics potential for discrimination and about legislation being enacted to stop such discrimination by insurers or employers, Wolf says the problem becomes more fundamental. She thinks that people begin to be seen as their genes and that their genetic medical records are perceived as "hidden truth." She calls this more insidious discrimination "geneticism.," which uses genetic testing and information to "create and reinforce power relationships in which some dominate and others are subordinated."

3. Insurance

Billings, Paul R.; Kohn, Mel A.; deCuevas, Margaret; Beckwith, Jonathan; Alper, Joseph S.; and Natowicz, Marvin R. **Discrimination as a Consequence of Genetic Testing.** *American Journal of Human Genetics* 50(3): 476-482, 1992.

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.

Brom, Marne E. **Insurers and Genetic Testing: Shopping for that Perfect Pair of Genes.** *Drake Law Review* 40(1): 121-148, 1990-1991.

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.

Cook, E. David. **Genetics and the British Insurance Industry.** *Journal of Medical Ethics* 1999 April; 25(2): 157-162.

After contrasting the British and U.S. insurance industries, the authors describe the Association of British Insurers voluntary moratorium on mandatory genetic testing and on weighted rate schedules based on genetic information.

Cowley, Geoffrey. **Flunk the Gene Test and Lose Your Insurance.** *Newsweek* 128(26): 48-50, 23 December 1996.

Describing a family who lost health insurance when one of their four children tested positive with a 'fragile X' gene, Cowley provides an overview of problems often connected with genetic testing: protecting privacy, loss of job or health insurance, or inability to adopt or be adopted.

Dickson, David. **UK Insurers Allowed to Use Genetic Tests.** *Nature* 6(11): 1199, November, 2000.

Noting that Britain is the first country in the world to authorize the use of genetic screening results to determine insurance premiums, Dickson discusses the response of consumer groups to this development and the possible impact of this policy for future genetic testing.

Frieden, Joyce. **Genetic Testing: What Will It Mean for Health Insurance?** *Business & Health* 9(3): 40-46, March 1991.

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.

Gevers, Sjef. **Use of Genetic Data, Employment and Insurance: An International Perspective.** *Bioethics* 7(2/3): 126-134, April 1993.

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.

Harper, Peter S. **Insurance and Genetic Testing**. *Lancet* 341(8839): 224-227, 23 January 1993. Letters. *Lancet* 341(8843): 495, 20 February 1993, and *Lancet* 341(8848): 833, 27 March 1993.

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.

Have You Had a Gene Test? [editorial]. *Lancet* 347(9885): 133, 20 January 1996. Comments. **Genetic Testing**. *Lancet* 347(9002): 685-686, 9 March 1996.

Noting establishment of the UK Health Department's Advisory Committee on Genetic Testing, the editors address problems they foresee involving the insurance industry, noting that ethical problems will be discussed frequently as new situations arise. They urge a consensus solution that information on a person's genes be restricted to "direct medical uses." Persons would "forgo any premium advantage in being able to show ...low risk." Four letters comment on the position.

Hoar, David I. **Implications of Preclinical Genetic Diagnosis**. In *Proceedings of the Annual Meeting of the American Council of Life Insurance*, pp. 147-155, 1990.

In a reprint of a speech given to the insurance industry, Hoar says that genetic 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."

Kapp, Marshall B. **Medicolegal, Employment, and Insurance Issues in APOE Genotyping and Alzheimer's Disease**. *Annals of the New York Academy of Science* 802: 139-148, 16 December 1996

Discussing the "extent to which medicolegal, employment and insurance principles developed in other genetic testing experience over the last decade" can be applied to Alzheimer's testing, he thinks that unique aspects of the new testing, now in a "rudimentary stage", "may compel modification" of principles now used.

Lapham, E. Virginia; Kozma, Chahira; and Weiss, Joan O. **Genetic Discrimination: Perspectives of Consumers**. *Science* 274(5287): 621-624, 25 October 1996.

As part of the Human Genome Education Model (HuGEM) Project of the Georgetown University Child Development Center and Alliance of Genetic Support Groups, 332 individuals were surveyed about their experiences with health insurance, life insurance, and employment. Of the respondents, 43% reported that they or members of their family have experienced genetic discrimination in one or more of the three areas. Since self-selection bias could apply, the authors suggest that a follow-up study be conducted using a random sampling of respondents from a clinic population.

Lowe, Robert. Genetic Testing and Insurance: Apocalypse Now? *Drake Law Review* 40(3): 507-532, 1991.

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.

National Institutes of Health (United States) - U.S. Department of Energy [NIH-DOE] Working Group on the Ethical, Legal and Social Implications (ELSI) of Human Genome Research. [Genetic Information and Health Insurance: Report of the Task Force on Genetic Information and Insurance](#). [Bethesda, MD]: NIH-DOE Program on ELSI, [1993]. 30 p. [http://www.nhgri.nih.gov:80/About_NHGRI/Der/Elsi/itf.html]

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."

Rothstein, Mark, ed. *Genetics and Life Insurance: Medical Underwriting and Social Policy*. Cambridge, MA: MIT Press, 2004. 293 p.

Noting that "...from a policy standpoint, we have yet to decide the degree to which genetic information of relevance in medical settings should be available for use in other settings", this comprehensive overview of genetics and life insurance contains a comparison of international policies on DNA and underwriting, a discussion of justice and genetic prediction, and an analysis of antitrust law as it applies to insurers' rate setting. A survey questionnaire on genetic testing and life insurance is included as an appendix.

U.S. Congress. Office of Technology Assessment. *Genetic Tests and Health Insurance: Results of a Survey--Background Paper*. Washington: U.S. Government Printing Office, October 1992. (OTA-BP-BA-98, S/N 052-003-01310-0.) 78 p.

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.

4. Forensics

Glasser, Ira. **Scientific Advances Carry a Moral Pricetag**. *Scientist* 5(11): 12, 27 May 1991.

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.

Hoeffel, Janet C. **The Dark Side of DNA Profiling: Unreliable Scientific Evidence Meets the Criminal Defendant**. *Stanford Law Review* 42(2): 465-538, January 1990.

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."

U.S. Congress. Office of Technology Assessment (OTA). [Genetic Witness: Forensic Uses of DNA Tests](#). Washington: U.S. Government Printing Office, July 1990. (OTA-BA-438, S/N 052-003-01203-1.) 196 p.

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.

5. Workplace/Environmental

American Medical Association. Council on Ethical and Judicial Affairs. **Use of Genetic Testing by Employers.** *JAMA Journal of the American Medical Association* 266(13): 1827-1830, 2 October 1991. Letters. *JAMA Journal of the American Medical Association* 267(9): 1207-1208, 4 March 1992.

The Council points out that employers, insurers, and law enforcement agencies all have uses for genetic information and techniques. It concludes that generally 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 be done without the informed consent of the employee or applicant for employment.

Bereano, Philip L. **DNA Identification Systems: Social Policy and Civil Liberties Concerns.** *International Journal of Bioethics* 3(1): 146-155, 1990.

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.

Billings, Paul; Rothstein, Mark A.; and Lippman, Abby. **But Is He Genetically Diseased? A Case Study.** *Hastings Center Report* 22(4): S18-S20, July-August 1992.

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.

Bohrer, Robert A. **Future Fall-Out from the Genetic Revolution.** *Futures* 24(7): 681-688, September 1992.

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.

Draper, Elaine. **Genetic Secrets: Social Issues of Medical Screening in a Genetic Age.** *Hastings Center Report* 22(4): S15-S18, July-August 1992.

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.

Hunt, Morton. **The Total Gene Screen.** *New York Times Magazine*: 33-34, 38, 50, 52, 55-57, 59-61, 19 January 1986.

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.

Strudler, Alan. **The Social Construction of Genetic Abnormality: Ethical Implications for Managerial Decisions in the Work Place.** *Journal of Business Ethics* 13(11): 839-848, November 1994.

Strudler looks at moral issues concerning a firm's use of genetic information about a prospective employee, reviewing literature on genetic abnormality and screening. HE concludes that there is a strong moral presumption against genetic screening in employment.

U.S. Congress. Office of Technology Assessment (OTA). [*Genetic Monitoring and Screening in the Workplace.*](#) Washington: U.S. Government Printing Office, 1990. (OTA-BA-455, S/N 052-003-01217-1.) 262 p.

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.

6. Counseling

Breo, Dennis L. **Altered Fates--Counseling Families With Inherited Breast Cancer.** *JAMA Journal of the American Medical Association* 269(15): 2017-2022, 21 April 1993.

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.

DeGrazia, David. **The Ethical Justification for Minimal Paternalism in the Use of the Predictive Test for Huntington's Disease.** *Journal of Clinical Ethics* 2(4): 219-228, Winter 1991.

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).

Elsas, Louis J. **A Clinical Approach to Legal and Ethical Problems in Human Genetics.** *Emory Law Journal* 39(3): 811-853, Summer 1990.

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.

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.

MacKay, Charles R. **The Physician as Fortune Teller: A Commentary on "The Ethical Justification for Minimal Paternalism."** *Journal of Clinical Ethics* 2(4): 228-238, Winter 1991.

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.)

Sosnowski, J. Richard. **Genetic Screening and Counseling: Ethical Considerations.** *Journal of the South Carolina Medical Association* 91(2): 73-78, February 1995.

Sosnowski provides a general medical ethics overview, and suggests ethical guidelines for physicians in the area of genetics: genetic screening is permissible, privacy is important, physicians should help establish guidelines for DNA databanks, and physicians should provide accurate information to patients.

Strong, Carson. **Tomorrow's Prenatal Genetic Testing: Should We Test for 'Minor' Diseases?** *Archives of Family Medicine* 2(11): 1187-1193, November 1993.

Strong advocates that "Physicians should honor requests for prenatal testing for diseases, including relatively minor ones, but not requests pertaining to nondisease characteristics." He presents other views, but finds each unsuitable. He holds that the "future role of reproductive genetic testing and counseling should be based on the imperfect but helpful, distinction between disease and nondisease."

U.S. Congress. Office of Technology Assessment. [*Cystic Fibrosis and DNA Tests: Implications of Carrier Screening.*](#) Washington: U.S. Government Printing Office, August 1992. (OTA-BA-532, S/N 052-003-01291-0.) 301 p. ***Genetic Counseling and Cystic Fibrosis Carrier Screening: Results of a Survey--Background Paper.*** Washington: U.S. Government Printing Office, September 1992. (OTA-BP-BA-97, S/N 052-003-01291-0.) 49 p. (See also, *JAMA Journal of the American Medical Association* 269 (15): 1921, 21 April 1993.)

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.

U.S. National Institutes of Health. [*Genetic Testing for Cystic Fibrosis*](#) NIH Consensus Statement Online 1997 Apr 14-16, 15(4): 1-37.

Wertz, Dorothy C., and Fletcher, John C., eds. ***Ethics and Human Genetics: A Cross-cultural Perspective.*** Berlin and New York: Springer-Verlag, 1989. 536 p.

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.

Whittaker, Lori A. **The Implications of the Human Genome Project for Family Practice.** *Journal of Family Practice* 35(3): 294-301, September 1992.

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.

7. Prenatal Diagnosis/Minors

American College of Medical Genetics. Newborn Screening Expert Group. **Newborn Screening: Toward a Uniform Screening Panel and System [Report for Public Comment].** Rockville, MD: U.S. Maternal and Child Health Bureau, March 2005. 329 p. Available online: <http://mchb.hrsa.gov/screening/>

In response to a 1999 request from the American Academy of Pediatrics for national newborn screening standards and policies, the Maternal and Child Health Bureau (MCHB) conducted a comprehensive review of the medical literature and surveyed experts in the field to develop recommendations. This analysis was used to delineate a minimum set of standards for state newborn screening programs, a resource allocation decision tree for testing options, and model policies and procedures for state MCHB offices to follow.

American Medical Association. Council on Ethical and Judicial Affairs. **Prenatal Screening.** *In its Code of Medical Ethics: Reports of the Council* 4(1): Report No. 44: 81-97, January 1993. [Also **Ethical Issues Related to Prenatal Genetic Testing.** *Archives of Family Medicine* 3(7): 633-642, July 1994.]

The report presents the "physician's role in promoting informed reproductive decisions and physician involvement in genetic selection and manipulation. In general, it would be ethically permissible to participate in genetic selection (abortion or embryo discard) or genetic manipulation to prevent, cure, or treat genetic disease. It would not be ethical to engage in selection on the basis of benign characteristics."

Bonnicksen, Andrea. **Genetic Diagnosis of Human Embryos.** *Hastings Center Report* 22(4): S5-S11, July-August 1992.

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.

Botkin, Jeffrey R. **Fetal Privacy and Confidentiality.** *Hastings Center Report* 25(5): 32-39, September-October 1995.

Botkin argues for "legal and ethical limitations on the application of prenatal testing and screening technology," saying that for "some medical conditions, respect for the privacy and confidentiality of the fetus outweigh parental rights to information about the fetus."

Clayton, Ellen Wright. **Genetic Testing in Children.** *Journal of Medicine and Philosophy* 22(3): 233-251, June 1997.

Clayton argues that there is room for substantial disagreement between parents and physicians about the appropriateness of genetic testing for children. She provides a series of cases, and concludes that there is room "to give increasing deference to the views of the child as the child grows older."

Cohen, Cynthia B. **Wrestling With the Future: Should We Test Children for Adult-onset Genetic Conditions?** *Kennedy Institute of Ethics Journal* 8(2): 111-130, June 1998.

In this special issue about genetic testing and genetic counseling, guest editor Cohen points out the reluctance to test children since genetics professionals often think the psychosocial harm is greater than any benefits, and the child's autonomy is violated in adulthood. The author thinks that testing should be considered utilizing many factors relevant to the particular child with the final decision making resting with the parents and the mature child.

Conway, S.P.; Allenby K.; and Pond, M.N. **Patient and Parental Attitudes Toward Genetic Screening and Its Implications at an Adult Cystic Fibrosis Centre.** *Clinical Genetics* 45(6): 308-312, June 1994.

The study examines cystic fibrosis patient/parent attitudes toward screening at one cystic fibrosis clinic, finding that 90%/88% support prenatal screening but that when questioned about terminating pregnancy the figures were 68%/84%. Few of the cystic fibrosis patients/parents thought screening should be limited to those with a prior history of the disease, and the authors say that those questioned conclude that "there should be the option of utilizing information available from genetic screening for cystic fibrosis to guide reproductive choices.

Hiller, Elaine H.; Landenburger, Gretchen; and Natowicz, Marvin R. **Public Participation in Medical Policy-Making and the Status of Consumer Autonomy: The Example of Newborn-Screening Programs in the United States.** *American Journal of Public Health* 87(8): 1280-1288, August 1997.

Saying that state newborn-screening programs are the largest group of genetic tests in the country, the authors surveyed them to look at public and parental participation. They conclude that increased public participation would "result in more representative policy-making and could enhance the quality of services provided by newborn-screening programs."

Holtzman, Neil A. **Genetic Screening and Public Health. [Editorial].** *American Journal of Public Health* 87(8): 1275-1276, August 1987.

Holtzman thinks that mandatory screening of newborns should include the opportunity for parents to decide on the merits of such genetic testing. Noting that parents rarely refuse testing when parental consent is required (Maryland), the author says that public health agencies have "a major responsibility to ensure the validity and utility of testing through adequate regulations."

Kristol, Elizabeth. **Picture Perfect: The Politics of Prenatal Testing.** *First Things* 32: 17-24, April 1993.

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.

Laberge, Claude M., and Knoppers, Bartha Maria. **Newborn Genetic Screening: Ethical and Social Considerations for the Nineties.** *International Journal of Bioethics* 2(1): 5-12, March 1991.

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.

Lippman, Abby. **Mother Matters: A Fresh Look at Prenatal Genetic Testing.** *Issues in Reproductive and Genetic Engineering* 5(2): 141-154, 1992.

Malinowski, Michael J. **Coming into Being: Law, Ethics, and the Practice of Prenatal Genetic Screening.** *Hastings Law Journal*: 45(6): 1435-1526, August 1994.

Malinowski's objectives are to present an "accurate portrayal of the practice of prenatal genetic screening; to analyze the opportunities it presents in the context of, and in contrast with, procreative liberty and abortion law; and to propose suggestions to ensure that the technology is welcomed, but with caution." He thinks that prenatal genetic screening is about "offering prospective parents difficult choices regarding the sacrifices they are willing to make to be parents...."

National Institutes of Health [NIH] (United States). **Consensus Development Conference. Genetic Testing for Cystic Fibrosis.** *Archives of Internal Medicine* 159(14): 1529-1539, 26 July 1999.

The Consensus Development Conference concluded that genetic testing for Cystic Fibrosis (CF) should be offered those with a family history of CF, partners of CF persons, couples planning a pregnancy and couples seeking prenatal care, but not to the general population or newborns. It urged further research and education, noting the need to protect privacy and to prevent discrimination or stigmatization.

Nolan, Kathleen. **First Fruits: Genetic Screening.** *Hastings Center Report* 22(4): S2-S4, July-August 1992.

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.

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.

Robertson, John A. **Ethical and Legal Issues in Preimplantation Genetic Screening.** *Fertility and Sterility* 57(1): 1-11, January 1992.

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.

Rothenberg, Karen H.; and Thomson, Elizabeth J., eds. **Women & Prenatal Testing: Facing the Challenges of Genetic Technology.** Columbus, OH: Ohio State University Press, 1994. 304 p.

The collection examines issues in reproductive genetic testing, particularly the psychological, sociocultural, ethical, legal and political applications. The editors point out that "Potentially, the major risk associated with reproductive genetic testing may come in not knowing how to cope with the information obtained from these procedures, rather than with the procedures themselves." Sixteen authors discuss testing from various perspectives such as choice, consent, justice, the parent-child relationship, accountability, law, and care.

Rothman, Barbara Katz. **Not All That Glitters Is Gold.** *Hastings Center Report* 22(4): S11-S15, July-August 1992.

Imagining a level of quality control in procreation that could make baby farms possible, Rothman warns of the dangers of preimplantation genetic diagnosis and the high physical, psychological, and social costs to the prospective mother.

Rothman, Barbara Katz. **The Tentative Pregnancy: How Amniocentesis Changes the Experience of Motherhood.** New York: W.W. Norton, 1993. 281 p.

Noting with irony that amniocentesis can not guarantee a "blue ribbon baby", Rothman describes the "shadow"

that reproductive technologies can cast over a pregnancy. Weighing the risks and benefits of such procedures, the author examines issues with commodification on one hand and the alleviation of great suffering on the other.

Wertz, Dorothy, D.; Fanos, Joanna H.; and Reilly, Philip R. **Genetic Testing for Children and Adolescents: Who Decides?** *JAMA Journal of the American Medical Association* 272(11): 875-881, 21 September 1994.

The authors write that genetic "testing may offer medical or psychological benefits, but harm parent-child bonds or the child's self concept." They review the legal status of minors as patients, their ability to make their own choices to assent, informed consent, ethical and legal requirements for competence and disclosure of genetic information, and offer guidelines for genetic testing of children.

8. Predisposition

American Society of Human Genetics. **Statement of The American Society of Human Genetics on Genetic Testing for Breast and Ovarian Cancer Predisposition.** *American Journal of Human Genetics* 55(5): i-iv, November 1994

Saying that BRCA1 may be responsible for about 5% of breast cancer cases, the Society says direct and reliable testing could be offered to members of families with strong breast-ovarian histories. Further research is recommended to determine optimal monitoring and prevention and public and professional education is needed to gain a "responsible approach to genetic testing." The Society notes that it is premature to offer population screening [1994].

Asch, Adrienne; and Geller, Gail. **Feminism, Bioethics, and Genetics.** In *Feminism & Bioethics: Beyond Reproduction*. Susan M. Wolf, ed. New York: Oxford University Press, 1996, pp. 318-350.

Saying that women are particularly affected by the proliferation of genetic information, Asch and Geller cover three areas: a background discussion of genetics and genetics research; feminist ethical analyses of various genetic testing with emphasis on autonomy, biological determinism, community, and women's moral reasoning; and thirdly, specific topics in genetics - presymptomatic breast cancer testing, prenatal diagnosis and provider-patient relations. There are 105 reference notes.

Baker, Stuart G.; and Freedman, Laurence S. **Potential Impact of Genetic Testing on Cancer Prevention Trials, Using Breast Cancer as an Example.** *Journal of the National Cancer Institute* 87(15): 1137-1144, 2 August 1995.

The authors conclude that highly predictive genetic testing for breast cancer will "provide impediments to prevention trials in the form of increased noncompliance", and selective trials that include only persons at high risk. They think that the "randomized trial will still provide the most reliable method of evaluating prevention strategies."

Burke, Wylie. **Taking Family History Seriously [Editorial].** *Annals of Internal Medicine* 143(5): 388-389, September 6, 2005. Available online: <http://www.annals.org/cgi/reprint/143/5/388.pdf>

Noting that the "...BRCA story illustrates the complexity inherent in the promise of genetically tailored health prevention", Burke describes the "tradeoffs and uncertainties" resulting from the U.S. Preventive Services Task Force (USPSTF) recommendations for BRCA mutation testing.

Carter, Michele A. **Patient-Provider Relationship in the Context of Genetic Testing for Hereditary Cancers.** *Journal of the National Cancer Institute Monograph* 17: 119-121, 1995.

Carter notes the speed of advancement in genetic testing, citing the BRCA1 gene linked with breast cancer, and considers issues important to the patient-provider relationship, how testing may alter this relationship, and ethical obligations inherent in the relationship.

Charo, R. Alta. **Legal and Regulatory Issues Surrounding Carrier Testing.** *Clinical Obstetrics and Gynecology* 36(93): 568-597, September 1993.

Charo, a member of the National Bioethics Advisory Commission, describes the public services and insurance coverage available for genetic screening and counseling; federal, state and private means of ensuring quality testing; the need to educate physicians about testing and providing a standard of genetic care; and federal and state protection against carrier discrimination.

DeVries, Bert B.A.; Mohkamsing, Serieta; van den Ouweland, Ans M.W.; et al. **Screening for the Fragile X Syndrome Among the Mentally Retarded.** *Journal of Medical Genetics* 36(6) 467-470, June 1999.

A screening program for the Fragile X Syndrome in the Netherlands studied 896 males and 685 females with an unknown cause for their mental retardation. Results indicated that clinical preselection for DNA testing in the mentally retarded is feasible by scoring on seven fragile X features.

Dickens, Bernard M.; Pei, Nancy; and Taylor, Kathryn M. **Legal and Ethical Issues in Genetic Testing and Counselling for Susceptibility to Breast, Ovarian and Colon Cancer.** *CMAJ Canadian Medical Association Journal* 154(6): 813-818, 15 March 1996.

The authors point out the legal distinction between breaking patient confidentiality and the "legitimate sharing of information in a patient's interest or to prevent harm to a third party," saying that health professionals have a duty to disclose sufficient information for informed decisions while safeguarding patient data.

Faden, Ruth R.; Tambor, Ellen S.; Chase, Gary A.; Geller, Gail; Hofman, Karen J.; and Holtzman, Neil A. **Attitudes of Physicians and Genetics Professionals Toward Cystic Fibrosis Carrier Screening.** *American Journal of Medical Genetics* 50(1): 1-11, March 1994.

A total of 1,140 primary care physicians and psychiatrists, and 280 medical geneticists and genetic counselors responded to a questionnaire surveying the attitudes of these professionals to population-based carrier testing for the cystic fibrosis gene. Only 43.9% of this group believed that such a test should be offered routinely, although 92% indicated that a couple who asked could be tested with a test that detected 80% of carriers. Those involved in genetic services were most opposed to routine screening based on the consequences of the 80% detection rate.

Guttmacher, Alan E.; Collins, Francis S.; and Carmona, Richard H. **The Family History—More Important Than Ever.** *New England Journal of Medicine* 351(22): 2333-6, November 25, 2004. Available online: <http://content.nejm.org/cgi/reprint/351/22/2333.pdf>

Celebrating the launch of a Web-based tool (<http://www.hhs.gov/familyhistory/>) designed to help people archive their medical records as part of the U.S. Surgeon General's Family History Initiative, the authors describe the Initiative as an advancement in "genetic literacy" that will work hand-in-hand with technical developments in genetic screening.

Haitt, Robert A. **Ethics and Economics of Genetic Testing for Breast Cancer.** Chapter 7 in *Reducing Breast Cancer Risk in Women*. Dordrecht, The Netherlands: Kluwer Academic Publishers, 1995, pp. 55-65.

The author notes the fears generated by breast cancer and says the interest in genetic testing will be high, generating "enormous new demands on the medical system." Ethical questions raised are the same as with other disease and include "individual rights, the potential conflict between individual rights and those of others, and the obligations and concerns of health care providers."

Hayes, Catherine V. **Genetic Testing for Huntington's Disease: A Family Issue.** *New England Journal of Medicine* 327(20): 1449-1451, 12 November 1992.

Humphries, S.E.; Galton, D.; and Nicholls, P. **Genetic Testing for Familial Hypercholesterolaemia: Practical and Ethical Issues.** *QJM* 90(3): 169-181, March 1997.

Commenting on the interaction of genetic components with environmental (e.g., diet or smoking), the authors note the difficulty of prediction in coronary artery disease (CAD), but they think that testing in familial hypercholesterolaemia is appropriate since early CAD is considerably increased. They comment on relevant ethical issues in the genetic testing of children and stress the need for family or children to give informed consent, noting the impact of increased anxiety.

Lancaster, Jonathan M.; Wiseman, Roger W.; and Berchuk, Andrew. **An Inevitable Dilemma: Prenatal Testing for Mutations in the BRCA1 Breast-Ovarian Cancer Susceptibility Gene.** *Obstetrics and Gynecology* 87(2): 306-309, February 1996.

Saying that the discovery of the BRCA1 susceptibility gene has created a demand for genetic testing, the authors point out that technical problems exist in developing widely available screening tests and applying them ethically. They state that patients and clinicians will have to "work together to determine a course of action with which they are most comfortable" since complex ethical, legal and social issues are involved.

Murray, Mary. **Nancy Wexler.** *New York Times Magazine*: 28-33, 13 February 1994.

Interviewing Nancy Wexler who has studied Huntington's disease for over 25 years and whose mother died of the disease, Murray reports that Wexler declines to say whether she has been tested. The author notes that in recent years many of the genes have been found for the estimated 4,000 inherited diseases but there is a "'painful hiatus' between prediction and prevention, Wexler warns is growing ever more perilous."

Parker, Lisa S. **Breast Cancer Genetic Screening and Critical Bioethics' Gaze.** *Journal of Medicine and Philosophy* 29(3): 313-337, June 1995.

Parker addresses and assesses the "neglect on the part of traditional bioethics" in developing protocols for breast cancer genetic screening, pointing out a role that bioethics should play.

Patenaude, Andrea Farkas. **Genetic Testing for Cancer: Psychological Approaches for Helping Patients and Families.** Washington, DC: American Psychological Association, 2005. 305 p.

Addressing the "emotional baggage" that accompanies decisions about genetic testing, Patenaude posits that "[t]herapists may help patients find deeper psychological acceptance of the realities and issues involved in genetic testing and may improve the likelihood that effective action will be taken and anxiety reduced." Chapters cover such topics as the clinical assessment of anxiety after disclosure of genetic testing results, prophylactic surgery for breast and ovarian cancers, family interactions and genetic testing, and a professional's duty to warn a client's family members if testing indicates the onset of a serious disease such as Huntington's.

Post, Stephen G.; Whitehouse, Peter J.; Binstock, Robert H., Bird, Thomas D.; et al. **The Clinical Introduction of Genetic Testing for Alzheimer Disease: An Ethical Perspective.** *JAMA Journal of the American Medical Association* 277(10): 832-836, 12 March 1997.

Twenty persons from the fields of genetics and public policy met several times to consider the clinical and ethical questions raised by genetic testing for Alzheimer Disease prediction or susceptibility in asymptomatic individuals or for diagnosis in persons with dementia. They concluded that "except for autosomal dominant early-onset families, genetic testing in asymptomatic individuals is unwarranted." Diagnosis of dementia "May prove useful but it remains under investigation." They conclude that premature introduction of testing should be avoided.

Post, Stephen G., and Whitehouse, Peter J., eds. **Genetic Testing for Alzheimer Disease: Ethical and Clinical Issues.** Johns Hopkins University Press, 1998. 284 p.

Divided into four parts, this collection provides an overview of genetic research on Alzheimer Disease (AD), describes the difficulties involved in genetic counseling for AD based on APOE alleles instead of single-gene

mutations, examines social issues such as genetic test patenting and actuarial policies for confidential data, and explores the public's perceptions and misperceptions about the nature and use of genetic testing.

Post, Stephen G. **On Not Jumping the Gun: Ethical Aspects of APOE Gene Testing for Alzheimer's Disease.** *Annals of the New York Academy of Science* 802: 111-119, 16 December 1996.

Post says that testing for Alzheimer's is "a premature introduction into the clinical context" and that it is necessary to address areas of "confidentiality, justice in access to genetic testing and testing guidelines."

Rothstein, Mark A. **Legal and Ethical Issues in the Laboratory Assessment of Genetic Susceptibility to Cancer.** *Birth Defects Original Article Series* 26(1): 175-190, 1990.

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 screening 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.

U.S. Centers for Disease Control and Prevention (CDC). **[Genetic Testing for Breast and Ovarian Cancer Susceptibility: Evaluating Direct-to-Consumer Marketing--Atlanta, Denver, Raleigh-Durham, and Seattle, 2003.](#)** *MMWR: Morbidity and Mortality Weekly Report* 2004 July 16; 53(27):603-6.

After a U.S. provider of BRCA testing (Myriad Genetic Laboratories, Inc., Salt Lake City, Utah) conducted a pilot direct-to-consumer (DTC) marketing campaign in two cities (Atlanta, Georgia, and Denver, Colorado), the CDC surveyed consumers and providers in the pilot sites and two comparison cities (Raleigh-Durham, North Carolina, and Seattle, Washington) to assess the impact of the campaign on consumer behaviors and health-care provider practices. The findings underscore the need for evidence-based recommendations for appropriate use of genetic tests and for the education of providers and the public to achieve maximum individual and public health benefit from genetic testing.

United States Preventive Services Task Force. **Genetic Risk Assessment and BRCA Mutation Testing for Breast and Ovarian Cancer Susceptibility: Recommendation Statement.** *Annals of Internal Medicine* 143(5): 355-361, September 6, 2005. Available online: <http://www.annals.org/cgi/reprint/143/5/355.pdf>

After a systematic review of the evidence relating to genetic screening for breast and ovarian cancer, the United States Preventive Services Task Force issued recommendations that only women with a family history of those cancers be referred for BRCA mutation testing. A one-page patient summary of these recommendations is available online at: <http://www.annals.org/cgi/reprint/143/5/I-47.pdf>

Wexler, Alice. **Mapping Fate: A Memoir of Family, Risk, and Genetic Research.** University of California Press, 1995. 321 p.

An historian, Wexler intertwines the stories of her mother's struggle with Huntington's disease, the discovery of a genetic marker for it, and the hope for a cure generated by this discovery. After analyzing the psychosocial issues raised by a predisposition to Huntington's disease in an era of feminism and patient's rights, the author describes the "toxic knowledge" that can result from genetic testing for a disease that has no cure.

1993, and it continues to be updated by NRCBL reference staff members Martina Darragh, Harriet Gray, Anita Nolen, and Susan Poland.

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