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Introduction to The Randolph W. Thrower Symposium: Genetics and the Law

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THE RANDOLPH W. THROWER SYMPOSIUM†

GENETICS AND THE LAW

INTRODUCTION

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Last year, Congress ushered us into a new era in genetics by initiating a fifteen-year study, the Human Genome Initiative. At the same time, the editors of the *Emory Law Journal* began their plans for this Symposium to ponder the legal issues inherent in this scientific endeavor.

The purpose of the Human Genome Initiative is to further our knowledge of genetics by gathering information about the 50,000 to 100,000 genes that make up the human genome. The emphasis will be on *mapping* genes (determining their location on the chromosomes) and *sequencing* genes (determining the identity of the constituent parts).

The Human Genome Initiative will not be starting from scratch, of course. About 1,500 genes have already been mapped and 600 have been sequenced.[1] Nevertheless, the entire project is of such a magnitude that one of the first orders of business will be the development of new technol-

† The Thrower Symposium is made possible by a gift from the family of Mr. Randolph W. Thrower, a distinguished alumnus of the Emory University School of Law. In previous years the Thrower series has featured a lecture presentation. This is the first year in which the series has featured a symposium, coordinated by the *Emory Law Journal*, at which participants were invited to present papers and commentary at a conference held at the Emory University School of Law on April 4, 1990.

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[1] McKusick, *Mapping and Sequencing the Human Genome*, 320 NEW ENG. J. MED. 910, 913 (1989).

ogies to speed the process of locating and identifying genes.[2] Researchers from other disciplines — such as computer science — will provide a necessary partnership in accomplishing project goals.[3] In the future, knowledge gleaned from the project will be used to develop genetic diagnostic and treatment technologies.[4]

The Emory Symposium had similar goals and used methodologies analogous to those of the Human Genome Initiative itself. The purpose of the Symposium was to sequence (identify) the policy issues inherent in medical genetics and to map them (determine their location in legal doctrine). It also built on previous research, much of it by Symposium participants.[5] The Symposium likewise used new technologies and aid from other disciplines (such as the use of deconstruction,[6] analysis of fiction,[7] the exploration of history,[8] and the use of surveys of geneticists[9]) to locate and identify the policy issues.

In the intensive day-long session at Emory University School of Law, the conference participants made great headway toward mapping and sequencing the policy issues in medical genetics. These issues fall into the categories of access, quality, confidentiality, decisionmaking control, and

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[2] OFFICE OF TECHNOLOGY ASSESSMENT, U.S. CONGRESS, MAPPING OUR GENES: GENOME PROJECTS: HOW BIG, HOW FAST? 7, 10, 12, 46-47 (1988).

[3] *Id.* at 47-48.

[4] *See, e.g.*, Friedman, *Progress Toward Human Gene Therapy*, 244 SCIENCE 1275 (1989); Hall, *James Watson and the Search for Biology's 'Holy Grail,'* SMITHSONIAN, Feb. 1990, at 41.

[5] *See, e.g.*, L. ANDREWS, MEDICAL GENETICS: A LEGAL FRONTIER (1987); GENETICS AND THE LAW (A. Milunsky & G. Annas eds. 1975); GENETICS AND THE LAW II (A. Milunsky & G. Annas eds. 1980); GENETICS AND THE LAW III (A. Milunsky & G. Annas eds. 1985); S. ELIAS & G. ANNAS, REPRODUCTIVE GENETICS AND THE LAW (1987); J. FLETCHER, COPING WITH GENETIC DISORDERS: A GUIDE FOR COUNSELING (1982); ETHICS AND HUMAN GENETICS: A CROSS-CULTURAL PERSPECTIVE (D. Wertz & J. Fletcher eds. 1989); GENETIC COUNSELING: FACTS, VALUES, AND NORMS (A. Capron, M. Lappé, R. Murray, T. Powledge, S. Twiss & D. Bergsma eds. 1979).

[6] Capron, *Which Ills to Bear?: Reevaluating the "Threat" of Modern Genetics*, 39 EMORY L.J. 665 (1990).

[7] Annas, *Mapping the Human Genome and the Meaning of Monster Mythology*, 39 EMORY L.J. 629 (1990).

[8] George Annas suggests that we explore the Nazi legacy and the United States' history of racism, sterilization, and immigration quotas to examine how economic concerns figure into issues of who should be allowed to reproduce. *Id.* at 645-46. Louis Elsas reminds us of American examples of abuses of patients in the name of science — at the Tuskegee Institute, the Willowbrook State School, and the Brooklyn Chronic Disease Hospital. Elsas, *A Clinical Approach to Legal and Ethical Problems in Human Genetics*, 39 EMORY L.J. 811, 830 (1990).

[9] Fletcher & Wertz, *Ethics, Law, and Medical Genetics: After the Human Genome is Mapped*, 39 EMORY L.J. 747 (1990).

self-concept.

Access

The first issue with respect to access is the development of incentives to ensure that appropriate genetic diagnostic and treatment technologies are developed.[10] The federal government has provided one incentive — funding for basic research into the genome. In this Symposium, Rebecca Eisenberg explores another potential incentive: the patent system.[11] She describes the circumstances under which a patent may be obtained and addresses the policy considerations regarding whether parts of the human genome should be patentable. The arguments for patent protection are that it provides potential financial rewards, which spurs researchers on, and that it makes public the invention so that others can use that knowledge for future scientific development. One argument against issuing patents in this area is that we may be paying twice for information about the human genome — once as taxpayers funding research and once as consumers paying royalties to the patent holder. Another argument is that, rather than encourage innovation, patent protection might stifle it since "disclosure through the patent system is likely to occur considerably later than disclosure motivated solely by scientific norms and rewards."[12]

To mediate between these different viewpoints on whether patent protection should be available, we need to analyze how closely the motivations of scientists working in this area resemble those of researchers in other areas. While patent protection may be necessary to assure that someone will build a better mousetrap, it might be less necessary in the genetics field, where the scientific issues may be more inherently interesting and more closely linked to other sorts of rewards (such as a Nobel Prize or tenure). Certainly much genetic research has occurred despite the fact that, as Eisenberg points out, some scientists assume patenting cannot be done and others suggest it should not be done.[13]

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[10] This step should take place after society decides that genetic technologies would be a useful way to spend our resources. See the discussion about resource allocation in Annas, *supra* note 7, at 643-44.

[11] Eisenberg, *Patenting the Human Genome*, 39 EMORY L.J. 721 (1990).

[12] *Id.* at 741.

[13] *Id.* at 738, 740-44.

Even if genetic information is uncovered or a genetic technology is developed in the first place, a second access issue arises regarding whether its use should be allowed. There should not be a presumption that just because a particular application of genetic technology is feasible, it should be used. John Fletcher and Dorothy Wertz, for example, argue against the use of prenatal diagnosis for sex selection purposes, unless it is related to a serious X-linked disorder.[14]

Already, there are some state laws that would prohibit the use of certain types of genetic technologies. For example, laws in seven states which ban research on conceptuses would appear to prohibit experimental gene therapy on embryos.[15] But such laws were not passed with gene therapy in mind. Rather, they were passed, for the most part, to prevent experimentation on later stage fetuses. The public policy question of whether certain genetic technologies are permissible should be addressed directly, so that their permissibility does not hinge on whether they happen to be covered by a broadly-worded, previously-enacted law.

A third aspect of access involves financial considerations. It is clear that even existing genetic technologies are not reaching all parts of the population equally. Women of higher socioeconomic status are more readily able to gain access to amniocentesis, for example. At the Symposium, John Fletcher suggested that the law should ensure that all people have access to at least a minimum of genetic services.

Quality

The law also has a potential role in assuring the quality of the genetic services and technologies that are provided.[16] This has several implications for the tests, treatments, and personnel that are used. Many previous genetic screening tests measured the level of a particular protein or enzyme

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[14] Fletcher & Wertz, *supra* note 9, at 789-90.

[15] LA. REV. STAT. ANN. § 9:122 (West Supp. 1989); ME. REV. STAT. ANN. tit. 22 § 1593 (1980); MASS. ANN. LAWS ch. 112 § 12J (Law. Co-op. 1985); MICH. COMP. LAWS ANN. § 333.2685 to .2692 (West 1980); N.D. CENT. CODE § 14-02.2-01 to -02 (Supp. 1989); R.I. GEN. LAWS § 11-54-1 (Supp. 1989); and UTAH CODE ANN. § 76-7-310 (1990). A recent decision held a similar Illinois law unconstitutional. *See* Lifchez v. Hartigan, 746 F. Supp. 1361 (N.D. Ill. 1990).

[16] For an example of the poor quality of genetic tests with respect to DNA fingerprinting in the criminal justice context, see Elsas, *supra* note 8, at 850-52.

in the body, the presence of which could be caused by one or more types of genetic defects. Stringent guidelines were suggested for the employment of these tests. They were to be designed to meet certain standards of sensitivity and specificity so that they would not result in an undue number of false positives or false negatives.[17] However, the types of tests that will be developed out of the Human Genome Initiative will involve direct assessment of the genes in situations in which there are variations in the type of genetic defects that cause a particular disorder. A test might be developed that is highly specific and sensitive to one particular defect causing the disorder, but would not pick up the other variations. Thus, from the standpoint of the individual or couple tested, there may be a large number of false negatives.

Such is the case currently with cystic fibrosis, for which the existing genetic test identifies only 70% of the carriers of the disease among couples who have not yet had an affected child.[18] This means that for each couple who is identified by the test, there will be about twenty-five more in limbo, in which one is a carrier and the other is not sure.[19] Serious questions are raised about whether a test should be employed when it leaves so many people in a state of uncertainty and worry.

Moreover, the quality of genetic diagnostic technologies cannot be judged on the narrow grounds of safety and efficacy, the usual Food and Drug Administration criteria. Because the information gained through genetic testing can have such enormous psychological, medical, legal, and financial ramifications, there needs to be an adequate system in place to assure that the appropriate level of counseling and education accompanies the use of a genetic test. Yet there are not enough genetic counselors available to meet the needs resulting from the increasing numbers of genetic tests. For instance, considering just the cystic fibrosis test, since one in twenty-five Caucasians is a carrier, eight million people would be identified by the test and would need follow-up counseling.[20] When the auxiliary services to allow a person to make the best possible use of a genetic test are not available, the results can be disastrous. Such was the case with

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[17] *See, e.g., Recommendations for Quality Assurance in Newborn Screening*, in LEGAL LIABILITIES AND QUALITY ASSURANCE IN NEWBORN SCREENING 63-65 (L. Andrews ed. 1985).

[18] Roberts, *To Test or Not to Test?*, 247 SCIENCE 17, 17 (1990).

[19] *Id.* at 18.

[20] *Id.* at n.19.

sickle cell disease testing, which was mandated by legislatures in the early 1970s without appropriate counseling opportunities.[21] In this Symposium, Louis Elsas provides a contemporary example of an eighteen-year-old woman who had been screened and learned that she was a carrier of Tay-Sachs.[22] She was psychologically traumatized by the results, in part because she had not been provided with sufficient information and counseling about the meaning of the test.

Genetic diagnostic technologies are becoming important to the medical care of an increasing number of individuals. Although initially obstetricians and gynecologists were the only health care professionals who were expected to routinely advise patients of relevant genetic tests, that duty now touches a variety of health care professionals. Genetic tests are available that indicate not a risk for the patient's offspring, but a risk for the patient himself or herself. Such tests provide the clinician information about an asymptomatic person's genetic predisposition to certain diseases (such as emphysema[23] or cardiovascular disease[24]) or the fact that the individual will suffer from a debilitating late-onset disorder such as Huntington's.[25] Currently, however, the teaching of genetics is not emphasized in medical schools. Physicians do not do very well in surveys of their knowledge of genetics.[26] Just as the law played a role in upgrading the genetic knowledge of obstetricians (by holding them liable in wrongful birth and wrongful life suits for inadequate genetic counseling and testing),[27] there may be malpractice suits in other areas of medicine to assure that physicians make appropriate use of genetic technologies.

Confidentiality

Genetic information is not only of value to the individual patient; em-

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[21] P. REILLY, GENETICS, LAW, AND SOCIAL POLICY 62-86 (1977).

[22] Elsas, *supra* note 8, at 839-42.

[23] *See, e.g.*, Calabrese, *Ecogenetics: Historical Foundation and Current Status*, 28 J. OCCUPATIONAL MED. 1096, 1098 (1986).

[24] Bishop, *New Medical Strategy Against Heart Disease Probes Inherited Flaws*, Wall St. J., Apr. 13, 1990, at 1.

[25] Gusella, Wexler, Conneally, Naylor, *et al.*, *A Polymorphic DNA Marker Genetically Linked to Huntington's Disease*, 306 NATURE 234 (1983).

[26] N.A. HOLTZMAN, PROCEED WITH CAUTION: PREDICTING GENETIC RISK IN THE RECOMBINANT DNA ERA 160-61 (1989).

[27] L. ANDREWS, *supra* note 5, at 135-48.

ployers, insurers, educational institutions, law enforcement officials, and others may wish to gain access to an individual's personal genetic profile. Yet there is a great potential for third parties to misuse genetic information. Employers, for example, might wish to exclude genetically-sensitive workers from jobs, rather than cleaning up the workplace to remove those toxins that present a risk of genetic damage or other health problems. Elsas points to a concrete example — an employer who wanted to institute screening for a genetic predisposition that put workers at risk of lung disease, while at the same time creating a workplace environment that increased risk of lung disease through the spread of mold.[28]

Revelation of genetic information can cause serious financial, emotional, and perhaps even physical harm to the patient. Geneticists themselves seem sensitive to this fact. When John Fletcher and Dorothy Wertz surveyed geneticists around the world they found very strong consensus that third parties, such as employers and insurers, should not have access to the results of the screening without the patient's consent.[29] An equally strong consensus emerged among Symposium participants that the law should protect the confidentiality of genetic information. The only situation in which an exception to that rule was proposed was when voluntary efforts to get the patient to contact a relative have failed, disclosure would help prevent serious imminent harm to the relative, and the relative would not readily find out about the genetic risk in another way.[30] Laws were also suggested to protect against genetic discrimination.[31]

Decisionmaking in the Genetics Context

Traditionally, patients choose medical services voluntarily, and on the basis of informed consent.[32] The Symposium dealt with whether that

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[28] Elsas, *supra* note 8, at 849-50.

[29] Fletcher, *Where in the World Are We Going With the New Genetics?*, 5 J. CONTEMP. HEALTH L. & POL'Y 33, 40 (1989).

[30] Fletcher & Wertz, *supra* note 9, at 788-89.

[31] *See, e.g.*, Capron, *supra* note 6, at 693.

[32] Patients need a great deal of information about genetic services in order to make appropriate choices. Louis Elsas suggests that the cursory interchange that is sufficient for gaining an informed consent in other areas of medicine is not appropriate with respect to genetic diagnostic and treatment technologies. Instead, he suggests that health care providers use the more comprehensive type of informed consent process that is required in situations in which the intervention being proposed is experimental.

model is appropriate with respect to genetics services as well. In fact, the question of whether genetic services should be voluntary or mandatory is so central to the range of policy issues inherent in the Human Genome Initiative that it was touched upon by six of the seven contributors to the Symposium.[33]

The issue of decisionmaking control is dealt with most extensively in John Robertson's article. He asks questions about the constitutional ramifications of private eugenics (in which parents use genetic information to choose whether to bear children) and public eugenics (in which the government requires carrier screening or prenatal screening).[34]

On the level of private eugenics, Robertson argues that people should be allowed to use prenatal genetic testing and to decide to abort based on the results of the testing. Yet Louis Elsas points out how complicated and difficult such a decision actually is, in describing the experience of a couple who chose to abort a fetus with a disorder that was treatable, when they felt the treatment required high emotional and financial costs.

On the level of public eugenics, lawmakers might advocate mandatory screening of *all* pregnant women, with the hopes that receiving information about the status of the fetus would cause women to abort a disabled fetus, thus saving the state the cost of providing services to an individual with a disability. Robertson does not go that far, but he does say, "Some persons will clamor loudly that individuals have a moral obligation to learn their carrier status and to avoid reproduction when there is a high risk that their offspring will have serious genetic disease."[35] Robertson argues that people do not have a right to impose those costs on others by, for example, bearing a child that will need financial support from society for its care.[36] However, financial concerns alone should not override an individual's right to refuse medical services. To quote Daniel Callahan: "Part of the very meaning of human community . . . entails a willingness of society to bear the social costs of individual freedom."[37]

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[33] *See* Annas, *supra* note 7, at 640-43; Capron, *supra* note 6, at 686-88; Elsas, *supra* note 8, 836-39; Fletcher & Wertz, *supra* note 9, at 764, 790; Robertson, *Procreative Liberty and Human Genetics*, 39 EMORY L.J. 697, 699, 708-09 (1990).

[34] Robertson, *supra* note 33, at 716-17.

[35] *Id.* at 717.

[36] *Id.* at 718.

[37] Callahan, *The Meaning and Significance of Genetic Disease: Philosophical Perspectives*, in

Concepts of Self

Whether voluntary or mandatory, genetic services can affect our sense of self and our relationship to others. George Annas and Alexander Capron raise the concern that as a result of the new genetics we may begin to think of people as just the sum of their genes. Annas senses that the combination of genetic diagnostic technologies and gene therapies may lead to dehumanization, where we manufacture people like products.[38]

The identification of a person as having a defect in a gene may cause the person to think of himself or herself as a defective person. As Elsas points out, the genetic defect is an integral part of self — unlike the infectious agent, which originates outside the self.[39] In addition, what is considered a permissible departure from normal will change as medical genetics can identify more and more genotypes. Along those lines, Elsas asks, "Will society stigmatize a survivor of widespread prenatal screening, such as a child with Down syndrome born to an older mother, as 'unfit to live'?"[40]

Capron identifies alterations that may occur in the relationship between parents and children. He notes that "the ability to treat genetic disease prenatally would alter what it means to be a 'good parent' and what is meant by 'defective' and 'normal,' as well as altering our sense of familial continuity."[41]

Future Mapping

The policy issues discussed so far make up a rough map of the questions that arise out of the Human Genome Initiative. There are many other questions that remain to be identified, however. Participants in the Symposium provide a variety of methodologies that could be used in that effort. Annas, for example, suggests several approaches. One is to concentrate on three categories of issues: those that affect the individual or fam-

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ETHICAL ISSUES IN HUMAN GENETICS: GENETIC COUNSELING AND THE USE OF GENETIC KNOWLEDGE 83, 89 (Fogarty International Proceedings No.13) (B. Hilton, D. Callahan, M. Harris, P. Condliffe & B. Berkley eds. 1973).

[38] Annas, *supra* note 7, at 648-49.

[39] Elsas, *supra* note 8, at 819.

[40] *Id.* at 832.

[41] Capron, *supra* note 6, at 677 n.28.

ily, those that affect society, and those that affect the species.[42] Annas also looks to fiction, drawing lessons from the works of Mary Shelley, H.G. Wells, William Shakespeare, Aldous Huxley, F. Scott Fitzgerald, and Robert Louis Stevenson in order to raise cautions about scientific attempts to understand and change the nature of human life and in order to identify particular risks that might need legal attention.

Symposium participants also suggested ways that the policy considerations can be handled — through a code of ethics for geneticists,[43] through advisory commissions,[44] through application of constitutional law[45] or common law principles,[46] and, if necessary, through moratoriums and bans.[47]

Since all of us have between four and ten genetic defects,[48] each of us has a stake in the policies that govern medical genetics. The choices that are made regarding access, quality, and confidentiality, and about mandatory versus voluntary genetic services will affect us all, and will shape the nature of the society in which we live.

At the Symposium, John Fletcher stressed the value of "prophetism" — predicting our future. But all six participants eloquently demonstrated how we can shape that future. The genetic map will be handed to us by the Human Genome Initiative, but it will be up to the law to show us where that map should lead.

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[42] Annas, *supra* note 7, at 639.

[43] Fletcher & Wertz, *supra* note 9, at 759-60.

[44] Annas, *supra* note 7, at 655-56; Capron, *supra* note 6, at 673-75.

[45] Robertson, *supra* note 31, at 713-15.

[46] Annas, *supra* note 7, at 657-58.

[47] *Id.* at 652-53.

[48] L. ANDREWS, NEW CONCEPTIONS: A CONSUMER'S GUIDE TO THE NEWEST INFERTILITY TREATMENTS, INCLUDING IN VITRO FERTILIZATION, ARTIFICIAL INSEMINATION, AND SURROGATE MOTHERHOOD 82 (1985).