

editors  
S. Armendarces  
R. Lisker

# HUMAN GENETICS

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PROCEEDINGS OF THE FIFTH INTERNATIONAL CONGRESS OF  
HUMAN GENETICS, MEXICO CITY, 10–15 OCTOBER 1976

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## **Lectures**



## THE LEGAL ASPECTS OF GENETIC SCREENING AND COUNSELING\*

MARGERY W. SHAW

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U.S.A.

It is both a challenge and a privilege for me to address an international audience on legal aspects of genetic screening and counseling even though my perspective is limited to American jurisprudence. Nevertheless, there are underlying principles of justice which have no national boundaries. These include the dignity of the individual, a sense of fairness, and the concept of equity. I hope that the case precedents and legal dicta cited here, although they are derived from American law, can be applied, in a broad sense, to all peoples of all nations.

I would also like to alert you to the fact that since I do not have a background in ethics my discussion will be limited to legal issues. However, I recognize that there is a great deal of overlap between legal and ethical problems. With these two caveats, I will proceed.

### Historical background

The interface between genetics and the law has a long and circuitous history. The Judaic code recognized the basic inheritance pattern of hemophilia many centuries before Mendel's discovery and exempted certain high risk males from circumcision.<sup>1</sup> Many primitive cultures had quasi-legal mechanisms for sanctioning infanticide of defective and malformed infants although the genetic etiology was unknown or not considered.<sup>2</sup> Taboos against incest are rooted in prehistory and remain alive today in the form of consanguinity statutes which prohibit marriage between close relatives.<sup>3</sup>

Following the rediscovery of Mendel's 'laws' in 1900, an ill-conceived eugenics movement flourished in the United States, giving rise to a variety of compulsory sterilization laws for feeble-mindedness, insanity, rape, epilepsy, habitual criminality and other categories of the 'hereditarily unfit'.<sup>4</sup> In 1924 a restrictive Immigration Act was passed by Congress based on conscious and unconscious racism and prejudice disguised as biological fact and genetic theory.<sup>5</sup>

In both the American sterilization laws and immigration laws, genetics was misused

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and distorted to serve political and social goals. We also remember vividly the Nazi extermination of European Jews under the guise of a negative eugenics program and Hitler's attempt to create a 'master race' by promoting a positive program of eugenics.<sup>6</sup>

More recently we have witnessed a misapplication of genetics and an attempt to infringe upon personal liberties in the ill-conceived sickle cell screening laws enacted by the U.S. Congress in 1972 and by at least 13 states.<sup>7</sup> With such a dismal record of legal and social policy issues applied to our knowledge of genetic principles it behooves us to proceed cautiously in making recommendations which may later carry the weight of legal authority.

### Legal aspects of programs aimed at the control of genetic disease

Modern laws of interest to geneticists raise many issues. The American legal system comes from three sources: common law (derived from the courts); statutory law (derived from the legislatures); and constitutional law. Four aspects of the law will be considered in the context of genetic screening, genetic counseling, and prenatal diagnosis. These are: the right of privacy; confidentiality of medical information; the doctrine of informed consent; and the tort of wrongful life.

#### *The right of privacy*

The right of privacy derives from a human need. This need, in turn, is undoubtedly rooted in our evolutionary development because there are many examples of territoriality drives in the animal world. Animals display a need for personal distance, social distance, and flight distance. There are distinct parallels between the territoriality rules of animal life and the trespass laws of human society: in each, there is a claim to private space.<sup>8</sup>

The legal concept of the right of privacy was enunciated in 1888 by Judge Cooley, who coined the phrase, 'the right to be let alone'.<sup>9</sup> This concept was emphasized in a law review article by Warren and Brandeis in 1890.<sup>10</sup>

Many other definitions of privacy have been offered by other authors but perhaps those most applicable to genetics and reproduction are 'freedom from unwanted intrusion', 'freedom from unwanted information about oneself in the hands of others', and 'a right to intimacy'.<sup>11</sup>

It is perhaps not a coincidence that our genitalia are colloquially called our 'private parts'. The constitutional right of privacy in America is centered around procreation. It may surprise you to learn that the U.S. Constitution does not mention the word 'privacy'. But procreational privacy in marriage was elevated to constitutional dimensions by the U.S. Supreme Court in 1965. In *Griswold v. Connecticut*<sup>12</sup>, a state statute which prohibited the use of contraceptives by married couples was struck down as unconstitutional. Justice Harlan said: 'I believe that the right of privacy in the marital relation is fundamental and basic — a personal right retained by the people.'



Seven years later, in 1972, the Court extended privacy in contraceptive use to unmarried persons. In *Eisenstadt v. Baird*<sup>13</sup> Justice Brennan said: 'If . . . the distribution of contraceptives to married persons cannot be prohibited, a ban on distribution to unmarried persons would be equally impermissible. . . If the right of privacy means anything it is the right of the *individual*, married or single, to . . . [decide] whether to bear or beget a child.'

Only 3 years ago, in 1973, the right of privacy took on added meaning in the *Roe v. Wade* abortion decision,<sup>14</sup> which held that the pregnant woman's right of privacy takes precedence over the right to life of the fetus until the stage of extrauterine viability. After that time the State may, if it chooses, proscribe abortion except to save the mother's life and health.

Only this year the U.S. Supreme Court expanded the right of the pregnant woman to control her own body by disallowing the requirement of spousal consent or parental consent in the case of minors for abortion.<sup>15</sup>

What do these 4 Supreme Court decisions mean to the genetic counselor? It can safely be stated that the counselor who chooses to do so may advise contraception and/or abortion as an alternative to reproduction for those couples who desire it. A much thornier issue, however, is whether the State has a right to prevent the birth of a genetically defective fetus. If the State has no compelling interest to intervene and protect the unborn, does it have an interest to interrupt a pregnancy not desired by society or antithetical to the general welfare? This question has not been addressed by American courts; I do not know if it has been heard in other countries. The right-to-life protagonists argue in favor of State intervention to protect all human life from the moment of conception, but not to prevent the life of an ill-conceived, genetically deformed fetus who is destined to a life of pain and misery or an early death. Any comprehensive program of genetic control would need to address this issue. Meanwhile, general legal guidelines would encompass the mother's right of privacy to decide if she wants to bear a genetically defective child.

We are witnessing a trend in American courts toward an extension of the right to privacy to include areas unrelated to procreation such as the school's invasion of privacy between parents and children<sup>16</sup> and the State's invasion of drug prescription records.<sup>17</sup> Of more interest to geneticists, perhaps, is the decision of the New Jersey Supreme Court in its holding on the Karen Anne Quinlan case based in part on the right of privacy.<sup>18</sup>

The court said: 'We think that the State's interest . . . weakens and the individual's right to privacy grows as the degree of bodily invasion increases and the prognosis dims. Ultimately there comes a point at which the individual's rights overcome the State interest [in keeping the patient alive].' This legal reasoning could easily be applied to extraordinary or heroic measures to keep alive genetically defective, malformed infants with no hope of cognitive or sapient life.

There are a number of other legal issues in addition to the right of privacy which arise with our developing technologies to control procreation. Although they will not be dealt with here they include legal questions surrounding artificial insemination, paternity exclusion, in vitro fertilization, cloning, adoption, fetal research, and gamete banks. The interested reader is referred to a recent book entitled *Genetics and the Law*.<sup>19</sup>

### *Confidentiality of medical information*

Confidentiality of medical information concerns the geneticist in several situations. First, there is the expectation of the patient that any personal information he or she divulges or that the physician discovers will be treated as confidential. Second, there is the dilemma of the genetic counselor who is concerned with the rights and needs of third parties, such as spouses or relatives, to be informed of genetic risks. And third, there is the rapid accumulation of genetic data such as the collection of pedigrees, genetic registries, and computerized data banks which poses special problems in keeping records confidential.

The Hippocratic oath includes a statement on the duty of the physician to maintain strict confidentiality<sup>20</sup>: 'What I may see or hear in the course of treatment in regard to the life of men, which on no account one must spread abroad, I will keep to myself, holding such things to be shameful to be spoken about.' Furthermore, the Principles of Medical Ethics of the American Medical Association<sup>21</sup> addresses the issue of doctor/patient confidentiality in the following terms: 'A physician may not reveal the confidence entrusted to him in the course of medical attendance, or the deficiencies he may observe in the character of patients unless he is required to do so by law or unless it becomes necessary in order to protect the welfare of the individual or of the community.'

These two statements form the cornerstone of medical confidentiality. But they are not absolute. There are times when the law recognizes that confidentiality may be breached to protect third parties. Several examples of case law illustrate not only the right, but the duty of a physician to breach a confidence. In *Pennison v. Provident Life and Accident Insurance Co.*<sup>22</sup> the court held that during marriage a husband has the right to obtain a full medical report from his wife's doctor. Thus, disclosure of medical records to a spouse would not be a violation of the right to privacy. Similarly, in *Curry v. Corn*,<sup>23</sup> a physician was not found liable to his patient for revealing medical information to the spouse. The court stated that during marriage each spouse has the right to know the existence of any disease or defect which may bear on the marital relation. Taken literally, this would protect the genetic counselor from liability for disclosure of genotypes or karyotypes to spouses which might have some bearing on the risks to their offspring or on potential sterility. These could include XXY, XO, translocations, sex-linked recessive heterozygosity in the female, autosomal dominant genes with late onset, or autosomal recessives in either spouse. Some situations might arise where illegitimacy is discovered and although the ethical or humane course of action might be to 'keep the secret' or even tell a lie, the law would probably protect disclosure.

Premarital genetic counseling raised the issue of disclosure to a potential spouse. The courts have not adjudicated this situation. However, there are cases in tort law where fraudulent misrepresentation to one's spouse before marriage (such as impotence or sterility) constitutes grounds for divorce or for nullifying the marriage.

In *Simonsen v. Swenson*,<sup>24</sup> the court held that where a physician discloses information which he believes is necessary to prevent the spread of infectious disease he cannot be held liable if he acts in good faith. Thus the absence of malice or the intent to do harm is recognized in law as a defense to disclosure. Other examples of a duty to warn

of infectious disease include negligent failure to disclose a diagnosis of TB, thereby putting the wife at risk,<sup>25</sup> negligence or fraud for failure to inform a patient's neighbor that smallpox is contagious,<sup>26</sup> and negligence in informing a family that typhoid fever<sup>27</sup> or scarlet fever<sup>28</sup> would not infect them or other family members. These cases suggest that relatives and spouses stand in a special relationship compared to other third parties and the counselor may have a special duty to disclose risks to them while maintaining confidentiality vis-a-vis the public at large.

A highly publicized case holding that a psychotherapist has a legal obligation to breach the confidentiality of a potentially dangerous patient was decided by the California Supreme Court only 3 months ago. In *Tarasoff v. Regents of the University of California*, the court held that the confidential character of professional communications must yield when disclosure is essential to avoid danger to others.<sup>29</sup> It stated: 'The protective privilege ends where the public peril begins.' In this case the 'public peril' involved just one person — the patient's girl friend — whom he confided he intended to kill and later did so. The psychologist (not a physician) was held to be under a legal duty to warn either the girl or her parents of his patient's threats. Failure to warn relatives and spouses of potential genetic risks might result in a cause of action in negligence against either physicians or non-physician genetic counselors.

The problem of third-party disclosure can be summarized by asking three questions: Do I have no right to disclose? Do I have a right to disclose, but no duty to do so? Do I have a duty to disclose? There are no simple answers to these questions. The law provides only general guidelines and the counselor must evaluate each situation with prudence and discretion.

Now, let us turn to the special problem of confidentiality of genetic and pedigree information when it has left the protected environment of the counselor's office note pads and file cabinets and is transferred to pooled records, statistical surveys, and computerized registries and data banks. Here the counselor has, in a sense, lost control of the information. As our society becomes more and more reliant on automated information storage and retrieval this problem will loom larger and larger. This applies not only to genetic information but to other kinds of personal data such as school records, tax returns, employment histories, and credit ratings. The data processing professionals assure me that there are foolproof methods of protecting the confidentiality of information stored in computers by rigid controls of access but this is little comfort when widespread abuse is known to occur. Legal deterrence can be accomplished by properly drawn statutes which provide protective measures and impose heavy penalties for misuse. Laws are now being written to protect the confidentiality of stored information. One of these is the Privacy Act of 1974 concerning federal records.<sup>30</sup> Also, state legislation in this area is proliferating.<sup>31</sup> It is incumbent on the geneticists who design registries to comply with both state and federal laws.

### *The doctrine of informed consent*

The law generally treats 2 individuals who enter into a contract on an equal basis and assumes that they will participate in arms-length bargaining to reach an agreement. But in