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The Human Genome Project's Implications for Autonomy, Respect, and Professionalism in Medical Genetics*

Roger B. Dworkin

Medical genetic practice has always fit uneasily into traditional models of the doctor-patient relationship and traditional autonomy-centered medical ethics analysis. The Human Genome Project thrusts the lack of fit into our consciousness by increasing the number of situations in which the traditional models simply make no sense or do not work. In this paper I shall take some early steps toward reformulating approaches to medical ethical analysis of issues posed by advances in genetics. After examining a few relevant cases I shall suggest that a focus on respect and professionalism will lead to sounder results than a focus on autonomy and existing professional relationships.

I. The Traditional Approach

The doctor-patient relationship establishes the basis for American medical law, and autonomy and nondirective counseling are the shibboleths of American genetic medical practice. The standard view of the doctor-patient relationship is that one doctor and one patient voluntarily enter into a relationship in which, in exchange for payment from or through the patient, the doctor assumes a fiduciary obligation to the patient. This is an obligation of utmost good faith and fair dealing. It is an obligation of exclusive loyalty in which the doctor must put the patient’s interests above all others.¹ The obligation is not only an obligation to provide reasonably

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Roger B. Dworkin

competent medical care, but also to maintain the patient's confidences learned during the existence of the relationship. The doctor may not divulge information about the patient to the patient's relatives, employers, insurers, or anybody else without the patient's permission, and s/he will be liable to the patient if s/he does so. Of course, there are exceptions to these obligations of single-minded loyalty and confidentiality. A doctor may violate confidentiality when the public interest supervenes, and occasionally danger to a single individual may suffice to permit, or even require, disclosure of private information. Nonetheless, the doctor-patient relationship establishes the usual parameters of medical liability. When a doctor wants to know whether to perform a test or a treatment on a competent adult patient, the doctor knows that (questions of payment aside) the patient's preferences are the only ones that matter. The doctor may not consider a spouse's request that a test not be performed if the patient wants the test. The relationship, as its name indicates, is between the doctor and the patient, not the doctor and some wider circle.

This focus on the individual patient is reflected in conventional views of medical ethics, which make patient autonomy the dominant value to be served, and by many legal doctrines that catapult individual patient decision making to a position of dominance. The most important of these legal doctrines, at least in theory, is the requirement that a physician obtain a patient's informed consent before performing any significant medical or surgical procedure upon the patient. The commitment to patient autonomy has its counterpart in the genetic counseling profession's commitment to nondirective counseling. Nondirective counseling is an approach to genetic counseling based on the premise that the role of the counselor is to present

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4 *Horne v. Patton*, op. cit. fn. 2.
7 The requirement of informed consent is frequently traced to a dictum by Judge Cardozo, "Every human being of adult years and sound mind has a right to determine what shall be done with his own body ..." *Schloendorff v. Society of New York Hospital*, 105 N.E. 92, 93 (N.Y. 1914). The two classic cases that expound the modern law of informed consent are *Canterbury v. Spence*, 464 F.2d 772 (D.C. Cir. 1972) and *Cobbs v. Grant*, 502 P.2d 1 (Cal. 1972). For a suggestion that the commitment to autonomy reflected in such cases is more rhetorical than real, see, Roger B. Dworkin, "Medical Law and Ethics in the Post-Autonomy Age," in 68 Ind. L.J. 727 (1993).
8 Clinical genetic services are provided by members of several professions, physicians, Ph.D. scientists, masters level genetic counselors and genetic associates, nurses, psychologists, and others. For the most part I mean the arguments developed in this article to apply regardless of the specific professional identity of the person(s) providing services. Unless the context suggests otherwise, terms like, "genetic counselor," or "genetic professional," in this article are meant to include anyone performing the relevant tasks.
II. The Inadequacy of Tradition

None of the traditional linchpins of medical law and ethics—the doctor-patient relationship, patient autonomy, and nondirective counseling—works very well in the genetic disease context. As the Human Genome Project and other genetics initiatives develop apace, the failings of the traditional approaches become more obvious and more important to correct.

The doctor-patient relationship is an unsound basis for genetic law and ethics because medical genetics makes no sense as an individual-centered branch of medicine. The *raison d'être* of genetic screening, testing, and counseling is to help *families* (and sometimes even larger groups), not individuals, deal with family diseases. People seek presymptomatic diagnosis of Huntington's Disease (HD), for which no treatment is presently available, in order to make decisions about whether they and *their mate* will attempt to reproduce. Surely, the decision whether to run a 50% risk of having a child with HD or to proceed without knowledge of whether that risk exists is a decision that affects both members of a couple, not only the one who may have the HD mutation. Learning whether one is a carrier of cystic fibrosis, sickle cell anemia, or any other recessive disease is nearly as important to one's mate as to oneself. Discovering that a boy has hemophilia is important to his mother, all her female relatives, and all their existing and potential sons. Discovering that a person has familial adenomatous polyposis of the colon (FAP) may be lifesaving information for that person's relatives. The family nature of genetic information requires rethinking rules that are based on a model of an individual uniquely affected by his broken leg or her pneumonia.

Similarly, what does it mean to talk about patient autonomy in the genetic disease context? As I have suggested elsewhere, autonomy is a term that hides multiple meanings. The two most important are liberal individualism, the idea that each individual is, and ought to be, free to make important decisions about him or herself; and physical essentialism, the idea that one's body is one's own and ought to be protected from interference by others. While each of these views has much to

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10 *Dworkin*, op. cit. fn. 7.
recommend it and serves us well in moderation,¹¹ neither seems an especially use-
ful guide to sound decision making about the ethical and social questions raised by
genetics.

A decision about or based on one person’s genetic health inevitably involves
more than that person. The patient’s spouse or other mate, parents, children, sib-
lings, and more distant relatives all have substantial stakes in the patient’s genetic
condition, stakes far greater than the ordinary stake that every person has in the
well-being of those who are close to him. Those persons have decisions to make
about their own health and reproduction that cannot be made intelligently without
information about their relative’s genetic health. To allow a patient to make deci-
sions without considering the interests of these other deeply involved persons is to
exalt the autonomy of one person at the expense of the autonomy of others.
Whether the net gain to autonomy will be greater from sharing or withholding in-
formation and from sharing or withholding decision-making power depends on the
particulars of the family and the genetic disease under consideration. Whatever the
impact on autonomy, the patient’s autonomy will have been served at the expense
of other values, such as doing good or preventing harm to others.

Similarly, to pretend that only one person’s body is involved in the genetic dis-
ease context, so that person has an absolute right to be let alone, is to ignore reality.
Shielding a person’s body from diagnosis of FAP may “shield” his children from
even knowing that they have a reason to seek diagnosis, monitoring, and even cure
for colon cancer. It is an extreme view of individual autonomy that would find that
result attractive.

None of this is to say that patients should be routinely submitted to tests or other
procedures against their will, or to suggest that the decisions of individual patients
are not entitled to great weight in the ethical practice of medical genetics. I mean
only to suggest that ethical analysis that considers other values and other people in
addition to the patient is likely to lead to better results more often than an autono-
my based approach.

Nondirectiveness is the handmaiden of autonomy.¹² Rooted attractively in a de-
sire to avoid repeating the horrors of the Holocaust and to respect different atti-
tudes and cultures,¹³ nondirectiveness exalts moral relativism to the level of dog-
ma. It assumes that untrained individuals confronted with often terrifying informa-

¹¹ For interesting development of the idea that one can recognize the importance of auton-
omy without elevating it to a status superior to all other values, see, Willard Gaylin and Bruce
Jennings, The Perversion of Autonomy, New York/London/Toronto/Sydney/Singapore:

¹² Beth A. Fine, “The Evolution of Nondirectiveness in Genetic Counseling and Implica-
Grandstrand Gervais, “Objectivity, Value Neutrality, and Nondirectiveness in Genetic Coun-
seling,” ibid., p. 119.

¹³ For a brief history of the development of the ethic of nondirectiveness, see, Fine, op.
cit. fn. 12, pp. 101 - 106.
tion can make life altering, often irreversible decisions without guidance from trained persons who are intimately familiar with the issues involved. In other words, in the name of respect for individuals, nondirectiveness abandons those individuals at the time they most need help. This makes no sense.

Directiveness and nondirectiveness are not fixed concepts. There is a range of directiveness that runs from none to the use of physical force. Answering questions about what the counselor would do or what other persons who have confronted the patient’s dilemma usually do, or explaining why the counselor thinks one course is better than another is a long way from physical force. Indeed, providing such information treats the patient as a person who deserves respect, engagement, support, and dialogue, rather than as an empty vessel to be filled with information and turned out to sea.

Moreover, even if nondirectiveness were as attractive as many persons seem to think, it is unattainable. Others have noted that tone of voice, body language, even what questions one chooses to pursue, all convey the counselor’s values. They convey the values, however, without opening them up to discussion. Thus, these subliminal cues to what the counselor thinks the patient should do may be more coercive than just telling the patient the counselor’s opinion.

In addition, it is easier to say one believes in nondirectiveness than to practice it. Counselors are human beings. How can they pretend they do not care about great moral issues? Is a counselor who refuses to provide prenatal diagnosis for sex choice behaving unethically? Does the answer depend upon what culture the person seeking the testing comes from? Or does ethical behavior require a counselor to refuse to be complicit in the abortion of healthy girls, a practice the counselor may abhor, because somebody else’s culture only values male children? Does ethical behavior require a counselor to remain silent and assist a couple to produce a child with a disability that the parents have or value without regard to the counselor’s own views or the impact on the potential child and on others? If the answer to that question is yes, it is surely an answer that cannot be reached without substantial analysis beyond mouthing the slogans of autonomy and nondirectiveness.

On the other hand, counselors, being human, are not above hubris and are not immune from the desire to assert power over others. These tendencies must be carefully guarded against in devising any sound approach to genetic practice. I shall argue below that a striking example of professional overweening is the widespread refusal to participate in presymptomatic genetic testing of children for late

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14 See, Gaylin and Jennings, op. cit. fn.11, pp. 153 - 176, discussing the various forms that “coercion” may take.

onset diseases in the absence of a medical benefit from early diagnosis. However, regardless of whether one shares that view, the general point remains valid: Genetic law and ethics must protect persons from unfounded claims of expertise and inappropriate assertions of power.

Thus, the problem is clear: We need to develop an ethic for genetic professionals (enforced when appropriate by law) that takes all relevant interests into account and that gives patients and others the full benefit of professional expertise, but that does not exalt the power and position of genetic professionals beyond the range of their professional competence.

III. The Relevance of the Human Genome Project

The problems I have identified with basing clinical genetic practice on the doctor-patient relationship, autonomy and nondirective counseling have always inhered in medical genetic practice. The Human Genome Project and its fallout have exacerbated the problems and made them more important to solve. As genetic practitioners become able to give definitive diagnoses to ever more persons, and as we learn that ever more conditions are genetic in origin, issues about professional loyalty, confidentiality, and the nature of professional genetic advice not only affect growing numbers of people, but they affect them in more definitive ways. In the not too distant past if a patient’s paternal grandfather had HD, all a genetic counselor could tell the patient was that his father had a 50% risk of developing the disease and that he had a 25% risk. The case for breaching the father’s privacy about his 50% risk was pretty weak. The information would only give the patient something to worry about, and maybe some not very clear guidance about reproductive decision making. It is a far different thing to be able to tell a person, as we can today, if his father has tested positive, that his father will develop HD and that therefore, he has a 50% chance of developing it too and that he too can be tested for it if he chooses. Now breaching the father’s confidentiality empowers the patient to learn specific information about himself and to act on that information. This makes the old question of whether to breach the father’s confidentiality newly pressing. Similarly, a young woman has a strong interest in knowing whether her mother’s breast cancer is genetic in origin. The question of whether to discuss a woman’s health with her daughter is not new, but science has changed the stakes, thereby suggesting that we should rethink the answer.

Modern developments also present opportunities to deal with people differently, depending on their genetic predispositions. This forces us to consider when differential treatment is acceptable, perhaps desirable, and when it constitutes invidious discrimination. Sloganeering for autonomy and nondirectiveness won’t answer these questions for us. Is it possible to devise a better analytical approach?
Announcing abstract principles to replace or supplement exclusive loyalty, autonomy, and nondirectiveness seems unlikely to lead to agreement or to an adaptable approach to the solution of future problems. Perhaps analyzing cases will prove more satisfactory and will provide a basis for inducing generally applicable principles.¹⁶

1. Twin Trailblazers

Bob and Tom are 29-year-old consanguineous twins. Their father manifested symptoms of HD at 35-years of age. The disease followed its usual course, and he died two years ago at age 50. Bob, who is married and the father of two children, is eager to learn his HD status in order to make financial plans for his family and to decide whether to have more children. Tom, who is engaged to Helen, does not want to know his status. Helen does not know that HD runs in Tom’s family; Tom is afraid she will leave him if he has the HD mutation and she finds out. He also is afraid that he “could not live” with the knowledge that he will develop the disease.

Remarkably, Bob and Tom are both employed as interstate bus drivers. They are both concerned about losing their jobs if their employer learns that they will develop HD, and they are also both worried about being unable to obtain health insurance for themselves and their existing and future children.

Bob appears at a university genetics clinic seeking presymptomatic diagnosis for HD. He asks the counselor what she will do about Tom and Helen, and he urges the counselor to promise to keep his HD status a secret from his employer and from all insurance companies. How ought the counselor to proceed?

Traditional analysis, based on the doctor-patient relationship, autonomy, and nondirectiveness would lead to the following conclusions: Bob is the patient; the counselor owes obligations only to him. She will test him and give him his diagnosis and prognosis. She will not deal with Tom or Helen and will not inform Bob’s employer or insurers of Bob’s status, regardless of what it is. The counselor will not discuss whether Bob has obligations to Tom, Helen, or anybody else and will offer no opinions about whether Bob should have more children, quit his job, or notify his employer if he has the HD mutation.

In my experience this is not what happens. Despite the rhetorical commitment to autonomy and nondirectiveness, counselors I know will urge Bob to think about

¹⁶ Two of the following cases are adaptations of cases that my co-authors and I analyzed in our book, David H. Smith, Kimberly A. Quaid, Roger B. Dworkin, Gregory P. Gramelspacher, Judith A. Granbois, and Gail H. Vance, Early Warning: Cases and Ethical Guidance for Presymptomatic Testing in Genetic Diseases, Bloomington / Indianapolis: Indiana University Press, 1998. My analysis of all three cases has benefitted enormously from the insights of those colleagues. However, they are not to be associated with my conclusions, some of which they would find appalling.
the impact his decision to be tested will have on Tom. They will involve Bob’s wife in the counseling process, but they will, in fact, refrain from offering an opinion about whether the couple should reproduce. If Bob tests positive for HD, they will worry about what to do about Bob’s employer. Morally, they will not find it easy to live with failing to do anything to try to prevent a bus crash. They will also worry about their legal liability regardless of whether they warn Bob’s employer or fail to do so. They will be angry that Bob may have trouble with his insurance company, and, therefore, they may urge Bob to buy insurance before he is tested or to otherwise “game the system” in order to avoid what they think is unjustified genetic discrimination. The heart of their commitment to nondirectiveness will be their unwillingness to tell Bob and his wife whether they should refrain from having children.

As the account of the case and my observation-based prediction of probable counselor behavior indicate, real cases are too rich and too difficult to be resolved by a wooden commitment to traditional values and relationships. Real counselor behavior suggests that counselors know that too. Unfortunately, their indoctrination into traditional values leaves them ill equipped to do more than intuit a better approach. What would a sound approach to this case entail?

Two good starting points would seem to be a focus on the number of persons with interests at stake and the importance of those interests, and on the professional competence of the counselor to respond to those persons’ needs. Interested persons include Bob, Bob’s wife, their children, arguably their future children, Tom, Helen, Bob’s and Tom’s employer, future passengers on their buses, other persons using the streets when Bob or Tom is driving, insurers, and other persons who are or would like to be insured by those companies.

In this case (although not in all) the interests of Bob, his wife, and their children do not seem to be in conflict. Bob wants to be tested in part to help his family unit, and nothing suggests that his wife or children have any other view. However, Tom’s interests are starkly opposed to Bob’s. Bob’s and Tom’s genetic status is the same. The test that reveals Bob’s status will also reveal Tom’s. For psychological and social reasons Tom does not want to know his status. The counselor can do some things that will be helpful to Tom if she is willing to recognize his claim to consideration and if she is willing to deviate from nondirectiveness.

It is not adequate for the counselor to fall back on the fact that her relationship is only with Bob. Tom’s concern is that he does not want to learn the information a genetic counselor can provide. Ironically, however, if the counselor ignores him

17 A broader range of persons – Tom’s future psychiatrist, the genetic counselors of unborn children, the minister who will or will not be called upon to marry Tom and Helen, countless others, and in a sense the whole society, which experiences the benefits and burdens of every person born with HD – is involved in a way. However, this observation only notes the extent to which no one is an island. One cannot allow analysis to be paralyzed while counting angels on the head of a pin.
because he has no relationship with her, Bob will learn his diagnosis and will almost surely communicate it directly or indirectly to Tom. Thus, ignoring Tom is likely to injure him in the very way he sought to avoid by refraining from entering into a relationship with a counselor. This catch-22 can best be avoided by the counselor adverting to Tom’s interests.

Of course, considering Tom’s interests does not mean that they must prevail. A conscientious counselor, who is, after all, in the business of providing, not withholding genetic information, will probably decide that Tom’s desire not to learn his diagnosis cannot prevent Bob from learning his. However, the counselor should deal with Bob in a directive way to make sure that he understands the importance of his brother not learning the diagnosis. She should tell Bob that it would be wrong for him to disclose his diagnosis to Tom. That will come closer to satisfying both brothers’ needs than refusing to test Bob or testing him and saying nothing about Tom. Autonomous actors decide what behavior to engage in after being instructed by countless persons—parents, teachers, clergy, club and civic leaders, politicians, the media, friends—about right and wrong. Telling Bob to keep his mouth shut does not deprive him of his autonomy. It increases his chance to autonomously choose to do the right thing. Nor does being directive involve the counselor in acting beyond her expertise. A genetic counselor involved in presymptomatic screening for HD will know how many persons prefer not to learn their status and what the impact on them of learning unwanted information is likely to be. Acting on this knowledge is quite appropriate.

The next question is what more, if anything, the counselor should do about Tom and Helen. Helen is more removed from the patient, Bob, than Tom is, and deciding how open communication between fiancees should be is not a matter of genetic counseling expertise. However, the counselor may have experience with the psychological wear and tear that persons at risk for HD experience, and she may have seen the impact that keeping such secrets has on relationships. Therefore, it would not be inappropriate for the counselor to ask Bob to tell Tom that the counselor is willing to meet with him to discuss his situation without revealing his diagnosis and to offer to refer Tom and Helen to a couple’s relationship counselor, someone who does have the relevant professional expertise.

The next question to arise involves what to do about Bob’s employment if his test result is positive. Here the law has some relevance. Nearly 25 years ago the California Supreme Court recognized that psychotherapists have a duty to make reasonable efforts to warn the intended victims of their homicidal patients.18 That decision has been widely adopted and expanded.19 If a counselor does nothing to reduce the likelihood of Bob having an HD-induced bus crash, the counselor may

18 Tarasoff, op. cit. fn. 5.
incur liability to the victims of that crash. However, that legal possibility does not tell us what the counselor’s ethical obligations are or even what actions the law requires the counselor to take to try to avoid a crash.

The simplest thing for the counselor to do is to abandon the commitment to non-directiveness. The counselor should explain to Bob that early symptoms of HD, which may escape detection, may impair his judgment and affect his physical movements enough to make it unsafe for Bob to drive a bus. She should then tell him what is obvious—that this creates an intolerable risk to innocent persons and that Bob should take steps to reduce that risk. It is quite reasonable to require Bob to come for frequent checkups at an expert facility (perhaps the genetics clinic, perhaps a specialized neurology clinic) and to tell him that failure to keep his appointments will lead the counselor to inform Bob’s boss of his condition. This approach gives Bob a chance to do the right thing while retaining his job as long as safety permits. It minimizes the risk to other people. If Bob fails to keep his appointments or if his checkups reveal the onset of symptoms, then the counselor is probably obligated to inform Bob’s employer unless Bob does so himself. This sacrifice of confidentiality is justified because it is likely to save lives. It is a false commitment to autonomy that would preserve Bob’s confidence at the risk of other people’s lives. Loss of life is the ultimate loss of autonomy.

Two more questions about highway safety remain. What should the counselor do about Tom’s employer, and how does a duty to make reasonable efforts to protect potential victims of a bus driver affect our views of what counselors should do about patients who simply drive cars? On one level Tom’s case is easy. The counselor has no relationship with Tom. Therefore, she has no duty to maintain his confidences to weigh against the demands of public safety. If Tom’s employer is readily identifiable, the counselor should inform the employer of the risk. However, this approach deviates from our earlier conclusion that the counselor should attempt to protect Tom’s desired ignorance of his diagnosis. Again this problem can be solved by encouraging Tom to come to the clinic. Even without presymptomatic testing he can be routinely monitored for symptoms. Once he shows symptoms of HD, his interest in remaining ignorant is gone, while the interest in safety and the duty to make reasonable efforts to warn remain. If Tom refuses to be seen, the counselor may be able to defer warning for some time by assuming that Tom’s age of onset will roughly correspond to Bob’s. Ultimately, however, no interest of a person in remaining uninformed about his own health can justify exposing dozens of persons to death or serious injury.

Of course, few people are interstate bus drivers. Almost everybody drives a car. If we say that Bob's counselor has a duty to make reasonable efforts to avoid an HD-caused bus crash, does it follow that every genetic counselor with a presymptomatic or early symptomatic HD patient must make reasonable efforts to avoid a car crash? The answer may well be yes, but the requirements of reasonableness may be small. Again, it is hard to see that nondirectiveness is very important here. Telling a patient to stop driving is a long, long way from practicing eugenics. What conceivable purpose could be served by refraining from urging the patient not to drive? Beyond that, however, little can be done. Telling an employer can effectively get a bus driver off the road. Even telling the Motor Vehicles Bureau is unlikely to keep a wilful car driver out from behind the wheel. Moreover, an automobile driver poses a danger to fewer people than a bus driver does. Directive counseling may be all that reasonableness requires.

Dealing with insurance companies is a different matter. Increasingly, both state and federal legislation is attempting to prevent discrimination based on genetic conditions or, at least, presymptomatic genetic conditions. For example, an Indiana statute prohibits insurance companies and HMOs from requiring applicants to submit to genetic screening or testing or to consider genetic screening or testing results in making coverage decisions or setting premiums. Federal insurance portability legislation prohibits group health plans and health insurers from treating presymptomatic genetic information as a pre-existing condition and also prohibits them from establishing eligibility rules based on genetic information. Thus, one could argue that a consensus is emerging that differential treatment by health insurers based on genetic status is inappropriate. If so, then one could also argue that there is nothing wrong with counselors trying to help patients circumvent inappropriate insurance company practices.

On the other hand, it seems clear that a counselor should not lie for a patient, if for no other reason than that establishing oneself as a liar is unlikely to promote a trusting relationship with the patient. Moreover, genetic counselors are unlikely to be experts about insurance, and they are even more unlikely to have thought through the policy implications of protecting their patients' access to health insurance. If insuring persons at risk for devastating genetic diseases will raise the cost of everybody's health insurance and prevent a large number of persons at the economic margin from being able to afford insurance, not everyone will agree that the right choice is to prefer the genetically disadvantaged over the economically disadvantaged. Surely, nothing in a genetic counselor's training equips her to make such choices. It is probably best for counselors to avoid discussion of insurance as beyond their competence.

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Thus, contrary to the dictates of the relationship-autonomy-nondirectiveness model, we have suggested that the counselor should (1) test Bob; (2) urge Bob to keep his diagnosis from Tom; (3) encourage Tom’s participation in the clinic; (4) offer a referral for couple’s counseling to Tom and Helen; (5) insist that Bob be checked often for symptoms; (6) alert Bob’s employer to Bob’s HD status if Bob refuses to cooperate, misses appointments, or refuses to inform the employer himself after becoming symptomatic; (7) alert Tom’s employer if it is reasonable to believe that Tom poses a hazard on the job; and (8) avoid discussing insurance. This approach maximizes respectful concern for everybody who is involved in any meaningful way, sacrifices almost nothing of real value, inflicts no harm on anyone, and avoids having genetic counselors deal with matters as to which they have no claim of professional expertise and as to which they are likely to make mistakes.

2. The Young

Carol, age 33, has undergone presymptomatic testing for HD, and has been diagnosed as carrying the mutation that will cause her to develop the disease. She now seeks presymptomatic testing for Dana, her 13-year-old daughter, and Edward, her 11-year-old son. While one might question whether Carol should have to give any reason for seeking such testing, she has explained her reasoning: She wants to know whether one or both of the children will develop HD so that she can plan for the children’s future. Carol plans to create a trust fund for each child. If both children are either HD positive or HD negative, she will provide the same amount of money for each. If, however, only one is positive, Carol will give that child’s trust 2/3 of the money and the HD negative child’s trust 1/3.

Dana is eager to be tested. She says, “Yeh, man, I saw Grandma die, and I’ve been having nightmares ever since. I want to know whether I’m gonna get this gross thing.” When asked whether he wants to be tested, Edward says, “I don’t care. Yeh, I guess so. Whatever Mom says.”

How should the genetic counselors respond to Carol’s request?

The majority view is that children should not be tested presymptomatically for conditions that have no treatment or cure. Most genetic professionals would refuse to test Dana and Edward even though one is eager and the other is willing to be tested.²³ What can justify this extreme departure from nondirectiveness?

The Human Genome Project's Implications

The reasoning of those who would refuse testing goes as follows: There is no cure or treatment for HD; therefore, no benefit can be derived from testing. 24 Many at-risk adults refuse presymptomatic testing for HD; 25 we should preserve the child's opportunity (or would they say "right"?) to make his or her own competent, adult decision. 27 A positive diagnosis may harm the child by having an adverse psychological effect on him or her, by causing the child to be "labeled," and by causing the parents to reject the child or otherwise deal with the child in a disadvantageous way. 28

All of these arguments exceed the proper bounds of professionalism. The focus on the absence of treatment or cure demonstrates the narrowness of the genetic professional's expertise. 29 Testing and diagnosis may have benefits, even if they have no medical benefits. A positive diagnosis may lead a family to make special economic provisions for the to-be-affected child as Carol wants to do here. It may lead them to direct the child into a career that may be entered quickly and pursued fairly long after symptoms appear. It may cause the family to shower the child with love, to take her to Disney World or the World Series. Countless benefits are imaginable. And, of course, 50% of the children tested will receive the incomparable benefit of a negative diagnosis and a life without fear of contracting or passing on HD.

It is true that many adults who are at risk for HD choose not to be tested. However, that is no reason to refrain from testing a child. Refusing to test so that the child can decide for him or herself during adulthood assumes that only testing, not refraining from testing, has an impact on the child. That simply is not true. Refusing to test forces the child to live throughout childhood and adolescence as a "person at risk for HD." This will inevitably become that child's self image, and may contribute to a decision not to be tested as an adult. Testing in adulthood would challenge the child's very perception of who s/he is. Those who refuse to test children label them and affect their lives just as fully as those who would test.


25 Ibid., p. 786.
26 Ibid.
27 Ibid., pp. 785, 786; Davis, op. cit. fn. 23, pp. 580 - 81.
29 Both the UK Working Party and the ASHG/ACMG report concede that testing may offer some benefits. Ibid., p. 786; ASHG/ACMG Report, op. cit. fn. 23. Nonetheless, both generally oppose testing in the absence of medical benefits.
The argument based on the negative effects of a positive test ignores the already mentioned 50% chance that the test will be negative and assumes that parents will do the wrong thing. Who are genetic professionals (alleged nonjudgmental champions of autonomy and nondirectiveness) to make such judgments? Some parents will behave badly; some will behave well. In the absence of negative information about particular parents, the judgment should not be one for the professional to make.

Parents can make all sorts of bad decisions for their children. Nonetheless, we remit medical decision making for children to their parents because we assume that parents know more than (minor) children, that they care more for their children than anybody else does, and that some entitlements should accompany the burdens of parenthood. Those arguments apply equally well in the genetic disease context as in any other. Most importantly, nothing suggests that genetic professionals are in any way better equipped than parents to make presymptomatic testing decisions for children. Thus, if Carol insists on the children being tested, I see no sound basis for refusing her request.

However, this is not to say that the center is legally obligated to test the children or that its personnel should just cheerfully go along with Carol’s decision if they disagree with it. Genetic counselors have no special insights into Carol’s family and no expertise about trust funds or other financial arrangements that families may make. However, they do have experience dealing with the impact of genetic diagnoses on families. If they have reason to believe that testing the children will do more harm than good, then they would be inappropriately depriving Carol of their expertise if they did not share their information and their concerns with her. They should try to talk her out of making a mistake. Directiveness is the way to serve autonomy. Refusing to test deprives Carol of her autonomy. Silently acquiescing in her desire for testing diminishes her autonomy by letting her make her decision in partial ignorance. Directive counseling without prohibition seems the soundest course.

One word of caution is in order. Counselors may have tested so few children that they really do not have expertise about the effects of doing so. If that is the case, directive counseling would be inappropriate. It would simply confound Carol’s de-

30 The ASHG/ACMG report simply assumes that the judgment is to be made by professionals while recognizing that the judgment is one about harms that are only potential: “Testing should be discouraged when the provider determines that potential harms of genetic testing in children and adolescents outweigh the potential benefits.” ASHG/ACMG Report, op. cit. fn. 23, pp. 1233 - 34. (Emphasis added).


cision making with the counselor’s prejudice. If the counselor’s position about pre-
symptomatic testing for HD is based on experience with other diseases or data
about such experience, then the counselor must be careful to present her opinion in
such a way as not to overstate the expert nature of her position.

3. Secrets and Lies

Mary’s brother died at age 6 of Tay Sachs Disease (TSD). Mary, who is now 23
and a client of a major genetics center, knows that she is a TSD carrier. Mary is
engaged to Jeff, who is 24, and who does not know that Mary ever had a brother.
Mary wants her counselor to arrange to have Jeff tested to determine whether he is
a TSD carrier, without informing him that Mary is a carrier. Her thinking is that if
Jeff is a carrier, she will marry him, have prenatal diagnosis without telling him
whenever she becomes pregnant, and abort affected fetuses while Jeff remains
blissfully ignorant. If Jeff is not a carrier, Mary thinks he has no reason to know
that she is. She thinks she can talk Jeff into being tested because he is an Ashkenazi
Jew, and he knows of the prevalence of TSD among members of that group. She
and can be re-tested or pretend to be tested and just tell Jeff that she is not a carrier.

A commitment to Mary’s autonomy, nondirectiveness, and an exclusive obliga-
tion to one’s patient would cause Mary’s genetic counselor to go along with this
scheme. Is that the ethical thing to do?

Pluralism is not relativism. Lying about matters of consequence is usually
wrong. Joining a profession does not disable one from following the dictates of
simple morality. Genetic counselors are not obligated to participate in every hare-
brained scheme their patients concoct as long as the scheme has some relationship
to genetics.

Moreover, if the genetic counselor begins to deal with Jeff, she will have estab-
lished a professional relationship with him. Therefore, she will owe obligations to
Jeff as well as to Mary. A focus on exclusive loyalty to one’s patient will not tell
the counselor what to do.

Mary’s genetic counselor should begin by getting directive with Mary. Jeff is a
human being who deserves to be treated with respect. He should have the fullest
available information in deciding whether to have carrier testing. He certainly
should not be deceived about his potential wife’s carrier status and excluded from
meaningful participation in decisions about whether to reproduce. A state is not
allowed to insist on spousal consent, or even notice, as a condition of abortion,33
but that does not mean that genetic counselors should participate in excluding hus-
bands from abortion decisions based on genetic indications. This is not a case in
which a woman is seeking abortion because an abusive husband makes it intoler-
able to bring a child into the relationship. This is a planned reproductive deception.

The counselor should explain to Mary that carrier detection for autosomal recessive diseases is primarily relevant for couples making reproductive decisions, and that, therefore, Jeff is entitled to the truth about Mary's carrier status, as well as his own. If Mary refuses to allow Jeff to be informed, then the counselor should refuse to arrange Jeff's carrier testing. The fact that Mary can lie and get somebody else to do it, does not change the counselor's obligations. The inability to create a perfect world does not exempt us from a duty to do the best we can.

Next the counselor should offer to refer Mary and Jeff to a couple's counselor. Although the genetic counselor is not an expert in relationship counseling, a person who lacks competence to deal with a situation may, nonetheless, recognize that a situation that needs to be dealt with exists. Here it is obvious that Mary's and Jeff's relationship leaves a great deal to be desired.

V. Respect and Professionalism

Several lessons emerge from these cases. First, they make it plain that many persons have interests to be protected in dealing with issues of genetic health. As human beings, each of these persons is entitled to be treated with respect. That means that as little as possible of each person's interests should be sacrificed. Considerable analysis is required to minimize the sacrifice of persons' interests.

First, when a claim is made that a particular course of conduct would injure or sacrifice an interest, that claim must be evaluated factually to see whether the interest really is being damaged in any significant way. For example, simply asserting that autonomy is sacrificed because something is done for a person is not helpful. It is more useful to examine the degree to which autonomy is actually lost. Once the real damage to an interest is assessed, that damage should be compared to the benefits gained or other losses avoided by making the sacrifice. Importantly, damage to an interest of one person may result in avoiding damage to the same interest of a different person. Restricting my autonomy may promote yours. When that is the case, the dilemma cannot be avoided simply by picking one person to prefer.

Even a high level of concern for autonomy cannot justify adherence to the practice of nondirective counseling. As we have seen, nondirective counseling often diminishes, rather than serves autonomy. In those instances it cannot be stubbornly adhered to. Moreover, even if autonomy will be sacrificed by a course of conduct, that can sometimes be justified as a way to protect the interests of other real people.

Treating persons with respect is not, or ought not to be understood as insisting on the dominance of autonomy. In real life we know that even if we do not always

admit it. For example, whatever one's views about assisted suicide for the terminally ill or hopelessly suffering, almost nobody is in favor of allowing persons upset about life's misfortunes to commit suicide unimpeded. If autonomy were the dominant value, then ethical behavior would require would-be rescuers to stand by and watch while unhappy persons jumped off bridges or blew their brains out. But, of course, persons who talk jumpers down or disarm would-be suicides are rightly celebrated as heroes, not criticized for behaving paternalistically. If autonomy were all that matters, then there would be no criticism of surrogate motherhood contracts on the grounds that they demean women by turning them into breeding machines.35 Such contracts would be universally celebrated as recognitions of women's autonomy.36 Prostitution, too, especially middle and upper class prostitution, by women who don't "need the money" would be accepted as the autonomous acts of free individuals. Needless to say, however, none of this comports with reality.

Respect for individuals requires considering all of a person's interests and attempting to do what is best for that person, all things considered. Autonomy and being free of even well-intentioned interference by others is an important interest to be considered along with all the others. That is why individual decisions are difficult. We can never say that a course should be pursued just because it is better than the alternative for a person's health. The question is whether the course is so much better that it is worth pursuing even at the sacrifice of the person's autonomy. With regard to preventing suicide, the general view is yes; with regard to surrogate motherhood, there is a split of opinion. With regard to genetic medicine questions will be extremely difficult to resolve.

If genetic medicine involved only single patients, then the model I am suggesting would look very much like the present fiduciary doctor-patient relationship, modified to reduce, but not eliminate, emphasis on informed consent and patient choice. The fact that genetic medicine involves multiple identifiable persons complicates the analysis. Respect for each individual requires that all of each person's interests be evaluated, and that decisions be made that will minimize damage to all the relevant interests, including the patient's interest in autonomy and in being treated as special by professionals with whom he has a relationship. This requires asking whether autonomy is really being sacrificed in a significant way as well as considering how important the values to be served by the sacrifice are. It requires searching for ways to make compromises that will provide second or third best solutions for everybody involved, rather than looking for the best solution for one person, unconcerned about what happens to everybody else. This is the kind of approach suggested in my discussion of the three cases above. One great value of the

35 Of course, there is a great deal of such criticism. For an analysis of surrogate motherhood and the criticisms of it and of the role of the law in regulating the practice see, Roger B. Dworkin, Limits: The Role of the Law in Bioethical Decision Making, Bloomington/Indianapolis: Indiana University Press, 1996, pp. 69 - 78.

36 As indeed they sometimes are. See, Judith T. Younger, "What the Baby M Case Is Really All About", in: 6 Law & Inequality 75 (1988).
cases is that they suggest that in concrete situations the suggested approach is not
impossibly difficult and that it leads to solutions that may commend themselves to
many who would not have been attracted to the analytical approach in the ab-
abstract.37

Serious dangers in the recommended approach are that professionals, freed of
the constraints of patient autonomy, exclusive loyalty, and nondirectiveness, may
ride roughshod over autonomy, seek to impose their own preferences on patients
and families, and even attempt to carry out eugenic policies. Therefore, respect for
individuals must be supplemented by attention to determining which individuals
are relevant and by adherence to the limits of professional expertise.

I have already noted that the number of persons theoretically affected by an in-
dividual's genetic health could be extended almost infinitely.38 Two dangers exist:
First, analysis could be stymied by the sheer number of persons whose interests
have to be considered. Second, refusing to draw lines may lead to the conclusion
that ethical behavior in any particular case will come to be understood as doing
what is best for the entire society. That will almost surely lead to eugenics as the
community as a whole will be understood to benefit from "survival of the fittest"
and the elimination of those who are a "drain" on the community.

The first danger is fairly easy to avoid. Only serious interests affected in major
ways should be considered. Everyone is affected in some way by everything, even
the mere existence of everybody else. We do not normally take de minimis impacts
into account in evaluating behavior, and there is no reason to do so in the genetic
disease context either. The interests of persons like those mentioned in note 17
above are simply too trivial, remote, and speculative to deserve consideration.

But what about the interests of the community? Surely some will argue that the
community would be better off if persons did not have costly and debilitating ge-
etic diseases. One needs no creative flair to imagine calls for mandatory genetic
testing, screening, counseling, and treatment, not to mention compulsory abortion
and sterilization. All of this could be proposed in the name of maximizing the
well-being of everybody who is relevant, especially, the ubiquitous taxpayers.
Avoiding eugenics requires two things: First, again, in considering interests we
must remember that the more direct and extreme an effect is on a person's interest,
the more that person's interest counts. That is why patients' interests will often
weigh more heavily than other persons' interests even after we have decided that
other persons' interests are relevant to determining what is sound behavior.
Second, it is essential to establish that in evaluating behavior the interests to be
considered are those of affected individuals with significant interests to protect,

37 This is a widely recognized advantage of casuistical analysis. See, e.g., Albert R. Jonsen
and Stephen Toulmin, The Abuse of Casuistry, Berkeley/Los Angeles/London: University
of California Press, 1988; Mark G. Kuczewski, Fragmentation and Consensus: Communitar-
The Human Genome Project's Implications

not those of the society. Generally, only identifiable persons may be considered. Avoiding concern for the unidentifiable will keep us off the slippery slope to eugenics.

The hardest cases, however, are those in which a group of persons, but not specific individuals, may be identified as having a major interest. A clear example is Bob’s and Tom’s passengers and other persons who could be injured by Bob’s or Tom’s dangerous driving. The fact that the interest at stake for such persons is life itself, an interest of the highest value, and that they are members of a small, definable group with its own membership criteria, not an amorphous, virtually universal group, like taxpayers, may be enough to justify considering those persons in deciding how to respond to the patient’s situation. Remember, however, that the contingent nature of the passengers’ interests led to the conclusion that the counselor should take a number of steps short of informing Bob’s and Tom’s employers to try to minimize damage to Bob and Tom. Considering a person’s or group’s interests does not mean they must always prevail.

Limiting the number of persons whose interests deserve consideration is not enough to protect patients from abuse. Genetic professionals occupy a position of power vis a vis their patients. The great virtue of nondirectiveness is that, if taken seriously, it avoids the imposition of a counselor’s views, which the power imbalance between counselor and patient would otherwise permit. The problems with nondirectiveness are that it cannot be achieved, and to the extent that it is achieved, it deprives patients of much needed guidance and expertise. I suggest that the way to solve this dilemma is to free genetic professionals from the constraint of nondirectiveness, while limiting them to areas of their professional expertise. A patient is entitled to a professional’s expertise. He is entitled not to be burdened with a professional’s nonprofessional opinions.

Obviously, it is difficult to define what falls within and without the scope of a professional’s expertise. Nonetheless, aids are available, and the effort is not impossible. Expertise comes from training and experience. Genetic professionals have professional expertise in matters about which they have received significant professional education and matters that they routinely observe and experience in their practice. They have expertise in genetics – inheritance patterns, risks, symptoms, prognoses, tests, available interventions; in the impact of genetic disease and carrier status on persons and families; in community resources available for referrals; in counseling techniques; and undoubtedly in other things as well. They do not have expertise in public policy; in economics; in insurance; in law; in ethics;39 in religion; in multi-cultural values and mores; in the dynamics of particular family relationships; and in many more things. Counselors should feel free, even obli-

39 This is true even if they have had some exposure to ethics in school. Professional expertise requires significant education, not just a course or two. Nobody would think that the average doctor is an expert in medical ethics or that the average lawyer is an expert in legal ethics even though courses in medical and legal ethics are ubiquitous.
gated to practice directive counseling in areas within their expertise, but should be ethically and perhaps legally constrained from offering advice in areas outside their expertise. To the extent that expertise is largely self-generated and is partly controlled by professional education, the profession itself can broaden the scope of its expertise, within limits allowed by licensure statutes, by broadening its education.

Limiting a profession to its area of expertise is hardly an unusual or an onerous restriction. Practicing a profession without a license is routinely prohibited. Courts and legislatures have trouble defining the limits of each profession, but they make the effort, and they succeed reasonably well. Some accountants would like to practice law, and some dentists would like to practice medicine, but both law and ethics prohibit the extension of practice beyond legitimate claims of expertise. The same kinds of limits are as appropriate for genetic professionals as they are for other professionals.

VI. Legal Implications

Much that I have recommended here is a matter of professional ethics. It can be accomplished by medical genetics professionals on their own and through their professional organizations. Genetic counselors can begin to practice directive counseling without any help from the law. However, the law can be helpful both to the profession and to patients and other relevant individuals in several ways.

First, the law should adapt current doctor-patient relationship law to make clear that duties related to genetic medicine do not run exclusively to patients. Genetic professionals deserve guidance and protection when they are deciding whether to breach confidentiality in order to warn a patient's relatives of their genetic risks, involve a spouse or mate or potential spouse or mate in reproductive decision making, and so forth. For the most part these protections should evolve slowly through common law litigation. That approach will provide less certainty than professionals may prefer. However, the real risks of liability are small. Moreover, it is impossible to define in the abstract the circumstances in which duties should be construed to run beyond the patient and the persons to whom those duties should run. Case by case analysis will minimize the costs of mistakes and even make it easy to undo the entire development of expanding duties if the expansion turns out to be ill advised. No pressing social need requires the entire focus to change immediately through legislation. Genetic professionals themselves can be effective in helping to shape the evolving law if their professional organizations participate constructively as amici curiae in litigation. Constructive participation, of course,

41 For an extended argument that common law adjudication should be viewed as presumptively the best legal response to bioethical dilemmas see, Dworkin, op. cit. fn. 35.
The Human Genome Project's Implications

means making a creative effort to contribute to an effective vision of genetic law for the 21st century, not simply opposing every potential imposition of liability on a genetic professional.

Second, professional licensure statutes and common law malpractice litigation together can be helpful in restricting genetic practitioners to their expertise. States should enforce their various unauthorized practice statutes to prevent genetic professionals who are not physicians, nurses, etc. from practicing professions for which they are not qualified. States should consider adopting licensure statutes for genetic counselors that define the practice of genetic counseling in a way that will make it plain that the practice of psychology, social work, etc. is not part of the job of a genetic counselor.

If genetic counselors engage in practices beyond their proper professional sphere, they should be liable for injuries they cause. Ordinarily violation of a licensure statute is irrelevant to the question of whether the unlicensed person behaved negligently, but a person who engages in another profession's activities will be held to the same standard of performance as a duly licensed person.

If a genetic professional acts beyond the scope of his or her expertise without violating an unauthorized practice statute, the lack of expertise should support a finding of negligence on which to base liability. For example, if a genetic counselor gives advice about how to deceive an insurance company, and the advice results in the patient being excluded from insurance coverage, the counselor's unreasonable behavior in acting beyond her expertise should give rise to liability for the damages the patient suffers from being rendered uninsurable.

The threats of malpractice liability and convictions for unauthorized practice will not get at every potential abuse. For example, they will be ineffective in forcing professionals to honor parents' requests to perform presymptomatic tests on their children. Nonetheless, if the profession itself adopts ethical standards that will restrict its members to their areas of expertise, those standards plus the use of malpractice litigation and licensure statutes where they can be effective should create a climate in which inexpert preferences are only infrequently imposed upon patients.

VII. Conclusion

A system based on exclusive loyalty to patients, patient autonomy, and nondirective counseling has never fit very well in the area of genetic medicine. Advances fostered by the human genome project make the lack of fit more obvious and more

43 Ibid.
44 See, Clayton, op. cit. fn. 32.
important than it used to be. Restructuring professional ethics so that they impose obligations to all relevant individuals (but not to society at large), focus on respect for individuals rather than patient autonomy, and seek to maximize the professional and minimize the nonprofessional contributions of genetic practitioners to their patients, will permit genetic professionals to maximize their usefulness, lead to increasingly sound ethical decision making for patients and other relevant individuals alike, and will avoid the dangers of imposition and eugenics. A small number of legal developments, particularly in the areas of professional licensure, expansion of duties, and professional negligence can help to promote these salutary ethical developments.

Zusammenfassung