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ETHICAL RESPONSIBILITIES OF PATIENTS AND CLINICAL GENETICISTS

ALLEN BUCHANAN, PH.D.*

I. NOT THE ETHICS OF PUBLIC POLICY

Ethical issues concerning genetic testing arise in two areas: public policy and individual conduct. Confusion results if these are not kept distinct because factors that may be relevant for determining what is acceptable public policy may not be relevant, or at least may not carry as much weight for determining the ethical, as opposed to the legal, responsibilities of individual patients and physicians. For example, even if there are strong reasons for concluding that a pregnant woman has an ethical obligation to undertake a genetic test that would detect and allow for the correction of a serious defect *in utero*, it does not follow that it would be ethically acceptable or prudent to sanction this obligation with the force of law. Concerns that a legal obligation might be enforced in a discriminatory way against poor women or racial minorities may argue against the imposition of a legal obligation.

Similarly, sound ethical analysis should recognize a distinction between types of public policy. Public policy includes not only the use of criminal, tort, or regulatory law to mandate, prohibit, or increase the costs of certain choices, but also extends to the provision of information and the use of techniques of persuasion through the "social marketing" of public health messages about the availability of genetic tests and the options available to those who test positive. A conclusive reason for not making it a legal duty for persons at risk to take a particular genetic test may not be a good reason against initiating a public awareness campaign designed to encourage the same group to take that test.

This essay focuses on the ethical responsibilities of individuals who must decide whether to undergo a genetic test or who have already received the results of such a test and on the responsibilities of

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the clinicians who reveal the test results to them.¹ The analysis offered here bears only indirectly, at best, on the ethics of public policy.

To state that the ethics of individual choice and the ethics of public policy are distinct is not to state that they are unrelated. In some cases, the existence of a particular public policy can shape the ethical responsibilities of individuals. For example, if public policy provides adequate health care for the elderly, then the ethical responsibilities of children for their aged parents may be narrower, or at least different in character, from what they would be in the absence of such policies. Similarly, public policy choices in the United States have resulted in a situation in which some people are at risk for uninsurability if they are known to have tested positive for a genetic condition,² a fact that may undercut what might otherwise be their obligation to inform their relatives that they too are at risk.

II. INDIVIDUALS' ETHICAL RESPONSIBILITIES: FIVE REASONS FOR A FRESH ANALYSIS CONCERNING TESTING AND TELLING

There are five reasons why a critical examination of the ethical responsibilities of individual patients and clinicians is necessary at this time.

1. *The Rapid Increase in the Number and Types of Genetic Tests*

First, a deluge of genetic tests is on the way. Suppose that the current repertoire of genetic tests is represented as a small stream of water pouring over a precipice and the existing totality of medical geneticists and genetic counselors as a single individual trying to catch the stream in a small cup. Now suppose that what had been a small stream becomes a huge waterfall — but that there is still only one individual, with one small cup, vainly attempting to catch the torrent.

It has been reported that one company plans to market a test kit whereby a single blood sample will be used to detect over 100 abnormal genetic conditions.³ A clinic at a state university medical center is already *direct* marketing a mail-in genetic test for hereditary hemochromatosis,⁴ and a mail-in genetic test for cystic fibrosis⁵ has

1. I will use the term "clinician" to cover both physicians and genetic counselors.

2. See Paul R. Billings et al., *Discrimination as a Consequence of Genetic Testing*, 50 AM. J. HUM. GENETICS 476, 478 (1992).

3. Electronic communication from Robert Cook-Deegan, Director of the National Cancer Policy Board, Institute of Medicine and Commission on Life Sciences, National Academy of Sciences (Oct. 1996).

4. Hereditary hemochromatosis is an autosomal recessive genetic disorder of excessive iron storage that can cause tissue damage and dysfunction of the liver, pancreas, heart,

already been marketed in the United Kingdom.⁶ In the coming decades, there will be many more tests with more ethical choices for more people, patients, and physicians.

2. *The Need to Challenge the Reproductive-Choice Paradigm*

Second, to a large extent, thinking about the ethical responsibilities of patients and clinicians has been shaped — and, I will argue, distorted — by the dominance of what I shall call “the reproductive-choice paradigm.”⁷ The reproductive-choice paradigm takes reproductive-predictive genetic tests to be the paradigm of genetic tests. Here “reproductive-predictive tests” means those genetic tests, such as those for cystic fibrosis carrier status, that prospective parents sometimes undergo to gain information to guide their reproductive decisions. Until very recently, by far the most common encounter between a patient and a clinician in which issues of genetic testing were broached focused on the question of whether to conceive or whether to carry a fetus to term. The existing consensus on the ethical responsibilities of physicians was forged as a response to the distinctive features of situations in which patients face *that* question. Among the most distinctive features of diagnostic-predictive tests are that (1) they touch upon the most intimate of human relationships for which the strongest case for privacy can be made; (2) the only options for responding to a positive test have been to accept the risk of a genetic disorder, to avoid conception, or to terminate the pregnancy, with the latter option raising one of the most divisive issues our society has known — the question of morality of abortion; and (3) it is in this area of reproductive choices that medical professionals are most acutely aware of the need to dissociate themselves from the taint of eugenics.⁸ All three of these facts lend support to the conclusion

and pituitary. See DORLAND'S MEDICAL DICTIONARY 747 (28th ed. 1994). A mail-in test exists to test for this genetic disorder and is offered by the Michigan State University DNA Diagnostic Program.

5. Cystic fibrosis (CF) is a generalized, autosomal recessive disorder of infants, children, and young adults, in which there is widespread dysfunction of the exocrine glands. See DORLAND'S MEDICAL DICTIONARY 628 (28th ed. 1994). CF is characterized by signs of chronic pulmonary disease due to excess mucus production in the respiratory tract. See *id.*

6. See Science and Technology Committee, House of Commons, Human Genetics: The Science and Its Consequences, VOL I, REP. AND MINUTES OF PROCEEDINGS, (3d Rep. 1994-95 session) [hereinafter SCIENCE AND TECHNOLOGY COMMITTEE].

7. See, e.g., Gail Geller et al., *Incorporation of Genetics in Primary Care Practice. Will Physicians Do the Counseling and Will They Be Directive?*, ARCHIVES FAMILY MED. 1119 (1993).

8. Eugenics is defined as the “branch of science concerned with the study of the hereditary improvement of man by genetic control.” MELLONI'S ILLUSTRATED MEDICAL DICTIONARY 156 (2d ed. 1985); see generally DANIEL J. KEVLES, IN THE NAME OF EUGENICS:

that physicians should avoid “imposing their own values” or the presumed values of society on patients in the reproductive-predictive test encounter.

To a large extent, it is in this distinctive context — the context that shapes the peculiar ethical significance of *reproductive-predictive genetic tests* — that a conception of the ethical responsibilities of clinicians has developed. But it should not be assumed that ethical principles for dealing with reproductive-predictive tests (such as amniocentesis to detect Down Syndrome, or a carrier test for cystic fibrosis) ought to be generalized to *all* genetic tests, including diagnostic-predictive tests (also called presymptomatic tests) for adult onset disorders such as breast cancer, colon cancer, and hereditary hemochromatosis. In particular, we should not assume that the same norm of nondirectiveness⁹ that may be appropriate for reproductive-predictive tests is also appropriate for diagnostic-predictive tests, and we should not assume that considerations of privacy weigh as heavily. Nor should we assume that all diagnostic-predictive tests are the same from an ethical point of view.

3. *The Ethical Responsibilities of Patients Regarding Genetic Testing*

It is crucial to focus on the ethical responsibilities of patients because there appears to be nothing approaching a consensus on what their ethical responsibilities are. The point is not that there are developed, but opposing, accounts of the ethical responsibilities of patients; rather, there is not a clear consensus that an account of the ethical responsibilities of patients is needed.

Here, too, the history of genetic testing — the dominance of the reproductive-choice paradigm — clouds the way we think about the ethical issues. Acceptance of the norm of nondirectiveness by genetic counselors, a norm which may be plausible for reproductive-predictive tests at a certain stage of social history, has encouraged a failure to develop an adequate conception of the ethical responsibilities of *patients*.

When clinicians observe the norm of nondirectiveness, but fail to make it clear that there is a distinction between what they as medical professionals should do and what the patients should do, clinicians may implicitly convey the message to patients that the choice confronting them is simply a matter of “personal values” — that the idea

GENETICS, AND THE USES OF HUMAN HEREDITY (1995) (identifying the past negative association of the word eugenics, after the second World War, with its connection to racism).

9. See *infra* text Section IV.E.

of ethical responsibility does not apply. But from the fact (if it is a fact) that clinicians should not tell patients what their ethical responsibilities are, it does not follow that patients have no ethical responsibilities, and that whatever they do is acceptable as long as it reflects their own values and is freely chosen. Ethical responsibilities, in general, are not reducible to acting consistently on whatever values one happens to have. Authenticity is an element of morality, not the whole of it. A person's action may be authentic in the sense that it is an expression of his deepest, stable values, yet it may be a wrongful action.

4. *The Need to Challenge the Therapeutic Pessimism Paradigm*

So far I have suggested that the fact that the formative context for the development of assumptions about the ethical responsibilities of patients and clinicians was the reproductive-predictive testing encounter encourages unfortunate generalizations to the ethics of diagnostic-predictive tests. I now want to suggest that the history of diagnostic-predictive tests, up to the present time, has the same tendency.

At present, with a few exceptions, diagnosis for genetic diseases outstrips treatment.¹⁰ This is especially true for the genetic tests that currently receive the most extensive media coverage and public discussion, including tests for the BRCA1¹¹ and BRCA2¹² genes, the APO E4¹³ Alzheimer's gene test, and the test for the Huntington's gene. In each of these cases, the medical benefit of testing is very dubious at present because there is no effective treatment for the condition. If all genetic tests were like these, the ethical landscape of genetic testing would be far simpler than it is in fact.

If we focus exclusively on tests for those late-onset conditions that are lethal and for which there is no effective treatment, such as Huntington's disease and Alzheimer's disease, we are likely to persist in the same ethical assumptions that developed in response to the reproductive-predictive test encounter. In particular, we will assume that

10. See TASK FORCE ON GENETIC TESTING, NIH-DOE WORKING GROUP ON ETHICAL, LEGAL, AND SOCIAL IMPLICATIONS OF HUMAN GENOME RESEARCH, PROMOTING SAFE AND EFFECTIVE GENETIC TESTING IN THE UNITED STATES 3 (1997).

11. BRCA1 is a gene believed to confer a greater than normal risk of breast cancer. See Jeffrey P. Struwing et al., *The Carrier Frequency of the BRCA1 185delAG Mutation is Approximately 1 Percent in Ashkenazi Jewish Individuals*, 11 NATURE GENETICS 198, 199 (1995).

12. BRCA2 is a genetic mutation that is believed to confer a higher than normal risk of breast cancer. See Karen H. Rothenberg, *Breast Cancer, The Genetic "Quick Fix" and the Jewish Community: Ethical, Legal, and Social Challenges*, 7 HEALTH MATRIX 97, 101 n.24 (1997).

13. "The inheritance of apolipoprotein (APO) E4 allele is an important risk factor for late-onset Alzheimer's disease." B. Permann et al., *Detection of Apolipoprotein E/dimeric Soluble Amyloid Beta Complexes in Alzheimer's Disease Brain Supernatants*, 240 BIOCHEM. BIOPHYS. RES. COMMUN. 715 (1997).

nondirectiveness is appropriate, at least in the situation where the patient has had the test and received a positive result. In such a case, observing the norm of nondirectiveness is easy, indeed virtually costless, because there is no treatment to refrain from recommending, and, therefore, no temptation to "impose one's values" by urging the patient to undergo a therapy one believes she should have.

The situation is quite different if there is an effective treatment for a potentially lethal disease that can be detected by a genetic test. In this case, the clinician will reasonably believe that there is a single right course of action, and that the ethical responsibilities of the patient are clear from the perspective of widely accepted and easily defended values.

At present there are few such conditions. Perhaps the clearest case of a lethal late-onset disease that meets this description is hereditary hemochromatosis.¹⁴ If detected early enough, hereditary hemochromatosis has a simple, inexpensive, virtually riskless, and fully effective treatment;¹⁵ yet this disease has devastating effects on the liver, heart, and endocrine system if left untreated.¹⁶ Genetically-based colon cancer is another example of a late-onset disease for which there is a genetic test and which can be successfully treated. However, the success rate for colon cancer treatment is lower and the treatment more invasive than that for hereditary hemochromatosis.¹⁷ It is reasonable to expect that in the future there will be more cases where those who test positive for a serious genetic condition will have the option of a successful treatment. For example, this is likely to be the case with at least some genetically-based cancers, once the role of the relevant genes in the development of the diseases is better understood.

The case of hereditary hemochromatosis, though at this time unusual so far as the efficacy of treatment goes, illustrates the danger of generalizing ethical assumptions that may make sense for diseases for which the prognosis is much bleaker. In the case of this relatively common genetic disease, a policy of nondirectiveness is much more dubious than in the case of reproductive-predictive tests or diagnostic-predictive tests for lethal, untreatable disorders such as Huntington's

14. See *supra* note 3.

15. The Treatment is "de-ironing" (removal of excess stored iron) by phlebotomies (removal of blood). See David L. Witte et al., *Hereditary Hemochromatosis*, 245 *CLINICA CHIMICA ACTA* 139, 143 (1996).

16. See *id.* at 139.

17. See Ian Tomlinson et al., *Molecular and Cellular Biology of Prostate Cancer*, 16 *CANCER & METASTASIS REVIEWS* 67-68 (1997).

disease, and it is much more difficult to dismiss the notion that patients have a responsibility to inform relatives that they are at risk. Moreover, when exploring the ethical responsibilities of clinicians¹⁸ it will become clear that the more a genetic disorder resembles the favorable case of hereditary hemochromatosis, the stronger the case is for clinicians to be "directive" by urging patients at risk to take the genetic test and for informing the patient who tests positive that he or she should tell relatives that they are at risk.

The fundamental point here is that it is necessary to think in terms of a continuum of tests for genetic disorders, with hereditary hemochromatosis at one end of the spectrum and Huntington's disease and Alzheimer's disease at the opposite extreme. On the left end of this spectrum, we have a disease that is cheaply, safely, and effectively treatable; on the right, we have those for which there presently is no effective treatment. Between the extremes we can locate the BRCA1 and BRCA2 gene tests (at present closer to the right end of the spectrum) and the test for the colon cancer susceptibility gene (closer to the left end). Depending upon where a test lies on the spectrum, different ethical judgments about the responsibilities of patients and clinical geneticists will apply.

5. *The Likelihood of Ethically Significant Changes in the Institutional Framework of Genetic Testing*

Fifth and finally, how we have come to think about the ethical responsibilities of patients and clinicians regarding genetic tests has also been shaped by features of the institutional framework within which decisions concerning testing have arisen. When assumptions about the institutional framework are treated as if they were permanent features of the ethical landscape, they become unquestioned framing assumptions for our thinking about ethical responsibilities. Although we tend to take their permanence for granted, they can change and, in some cases, already are changing.

There are two basic institutional facts that have framed our thinking about the ethics of telling and testing: (1) a positive test for a genetic condition carries the risk of insurance discrimination,¹⁹ and (2) genetic testing is a relatively rare event, not an integral element of

18. See *infra* text Section IV.

19. See Billings et al., *supra* note 2, at 481 (concluding that many health care institutions erroneously perceive genetic conditions as "extremely serious," "disabling," or "lethal," and as a result of this misconception, insurance institutions and employers use genetic testing information inappropriately by restricting or limiting access to public entitlements such as health care or employment).

routine health care, and physicians play a pivotal role in broaching the issue of whether to have a genetic test and how to respond to the results.

The actual extent of insurance discrimination on genetic grounds is disputable. It is quite possible that anecdotal evidence of discrimination overestimates the frequency with which individuals are denied insurance because they are known to have a genetic condition.²⁰ More importantly, the risk exists only for insurance policies whose issuance is conditional on medical underwriting, and most Americans who have private insurance get it through large group policies in which there is no medical underwriting.²¹ Nevertheless, the public perception seems to be that insurance discrimination on genetic grounds is or, with the emergence of more genetic tests, will become, a major problem.²² This perception, whether accurate or not, has led to a flurry of legislative initiatives, at the state and federal levels, that may in time succeed in prohibiting insurance discrimination on genetic grounds.²³

Whether we assume that there is a risk of insurance discrimination, and how serious we assume that risk to be, makes a difference when determining the ethical responsibilities of both patients and physicians. For this reason it is important to note that this first institutional fact may change, and to explore the difference this would make in how we should understand the ethical responsibilities of clinicians and patients (as well as individuals who may not be the patient of any physician). The change could occur either through legislation prohibiting refusal to insure on genetic grounds²⁴ or, less probably, through the replacement of the competitive private health care insurance market with a universal coverage, single-payer system.

The second institutional fact has two parts: (1) the relatively small role that genetic testing plays in health care at this time (especially outside the reproductive-choice context) and (2) the fact that when genetic testing does occur the physician controls access to these tests.

20. See *id.* at 477.

21. See Joel Cantor et al., *Private Employment-Based Health Insurance in Ten States*, 14 HEALTH AFFAIRS 199, 205 (1995).

22. See Karen H. Rothenberg, *Genetic Information and Health Insurance: State Legislative Approaches*, 23 J.L. MED. & ETHICS 312 (1995).

23. See Rothenberg, *supra* note 22, at 313-17; see, e.g., H.R. 306, 105th Cong. (1997) (prohibiting discrimination against individuals and their family members on the basis of genetic information, or a request for services); H.R. 328, 105th Cong. (1997); S. 89, 105th Cong. (1997); S. 1600, 104th Cong. (1996); S. 1416, 104th Cong. (1995); H.R. 2690, 104th Cong. (1995);

24. See Rothenberg, *supra* note 22 and accompanying text.

This second feature is already changing. As noted earlier, companies in the United States and the United Kingdom are already “direct marketing” home genetic test kits²⁵ which require no role for a physician. There is every reason to believe that this trend will grow with the expanding repertoire of genetic tests.

It is difficult at present to know whether, or when, the use of diagnostic-predictive genetic tests will become pervasive in the United States’ health care market. Much will depend upon whether the current trend toward managed care continues (which seems likely) and on the willingness of managed care organizations to include genetic tests among the services they cover.²⁶ If inexpensive, “one-shot” blood or cheek cell sample genetic tests that detect dozens of genetic conditions at a time become available, and if the problem of insurance discrimination is solved, genetic testing may become a routine part of health care. If genetic testing does become a routine part of health care, it will no longer be the case that unless individuals who tests positive for a genetic condition inform relatives of the results of their tests, they will be unlikely to know that they are at risk. This point is worth reflecting upon, because it is usually assumed that the “age of genetic medicine” will be one in which the crucial and recurrent issue for patients who test positive will be: What are my obligations, if any, to inform relatives at risk? If everyone is getting a standard battery of genetic tests, this question will lose much of its urgency.

The time is ripe, then, for a critical investigation of the ethics of genetic “testing and telling” for patients and physicians. In particular, it is important to avoid begging important questions by assuming that all genetic tests or even all genetic tests of a general type, such as diagnostic-predictive tests, are alike from an ethical point of view. Likewise, we must be careful to understand the role which institutional framework assumptions are playing in our beliefs about what our ethical responsibilities are.

To correct what I take to be the distorting focus on reproductive-predictive tests, I will concentrate on diagnostic-predictive tests, but much of what I say will apply to reproductive-predictive tests as well. At relevant points in the investigation, I will ask how changing our

25. See SCIENCE AND TECHNOLOGY COMMITTEE, *supra* note 6 and accompanying text.

26. Until recently it was assumed that health maintenance organizations (HMOs) would encourage preventive care. If this assumption holds, then one might predict that HMOs would be quick to utilize at least those diagnostic-predictive genetic tests that detect diseases that are responsive to therapies or life-style changes. But if enrollments are unstable — if individuals shift from one plan to another over time — the incentive for prevention is diminished.

assumptions about institutional arrangements affects ethical conclusions.

III. PATIENTS' ETHICAL RESPONSIBILITIES

There are two decision points at which the question of ethical responsibilities for patients arises: (1) deciding whether to have a genetic test (or whether to inquire about having one) and (2) deciding what to do in response to receiving the results of the test.²⁷

This section will concentrate on the latter, but much of what is written will have evident parallels for the former. The subject of this inquiry is even narrower. That is, what ethical responsibilities to "tell" — to inform others of the positive results of the test — might the patient have and to whom?

For example, John has just received a positive result for a diagnostic-predictive genetic test. Does John have a responsibility to inform relatives that they are at risk for the condition? If so, *which* relatives (e.g., second and third cousins, or just first cousins, or only relatives of the first degree) and *what constitutes fulfillment of this responsibility* (e.g., how much time and effort must he expend to contact the required relatives he should contact and to explain the significance of the test in terms they can understand)?

To answer these questions two things are necessary. First, we must clarify the possible *sources* of duties to tell, the moral considerations that support the assertion that there is a duty; second, we must articulate the factors that determine the *scope* and *weight* of whatever duties there are and see whether, or to what extent, these factors are present in various types of test situations, that is, for tests for various types of conditions.

A. *Possible Sources of Duties to Tell: General and Special Obligations to Prevent Harm*

Virtually all known ethical views, whether ancient or modern, religious or secular, recognize a general obligation not to cause harm to innocent persons. Most also recognize a general obligation to prevent harm as well, at least if the harm is serious, even if this is understood to be a more circumscribed obligation than the obligation not to cause harm. Thus for example, one might be morally required to risk

27. For brevity I will continue to use the term "patient" to distinguish these individuals from physicians, while acknowledging that the prospect of home-test kits means that individuals can be tested without being patients.

one's life to save an innocent person one has caused to be in peril, but not to save someone for whose endangerment one did not cause.

To say that an obligation is general is to say that it applies to all individuals, not simply those who stand in certain special relationships to one another (e.g., parent-child, physician-patient). General ethical obligations are those that are thought to provide the basic moral framework for interactions among persons.

To say that there is a general obligation to prevent harm, and not merely avoid causing harm, is not to deny, of course, that the distinction between causing harm and not preventing harm is morally significant in many contexts. More blame may attach to causing harm, and the costs an agent is expected to bear in order to avoid causing harm may be greater than those for preventing harm that one does not cause. However, the same basic concern for human well-being that ultimately grounds the obligation not to cause harm, grounds at least a presumption that one ought to prevent harm to persons as well. Generally speaking, whether an individual may bear some responsibility for a harm depends upon whether he was in a position of control, that is, whether he had the ability to prevent the harm, not whether he caused the harm. There are a number of reasons for believing that the obligation to prevent harm is far from an unconditional one. The most important, perhaps, is that if persons were expected to prevent all the harms they were capable of preventing they would have few resources and energy left for their own projects and for showing special regard for those with whom they are intimately connected, such as members of their own families. An open-ended obligation to prevent harm would be absurdly onerous; it would make us slaves to the risky behavior of others. Therefore, the scope of the obligation to prevent harm, whatever its weight *vis-à-vis* other obligations with which it may conflict on occasion, must be understood as being limited by a "reasonable cost" clause. The costs one ought to bear in order to prevent harm one does not cause are generally lower than those one ought to bear to avoid causing harm.

Other things being equal, what constitutes reasonable costs depends, in part, upon the seriousness of the harm to be prevented. Thus, although I may not be obligated to risk my life to rescue a stranger from a burning building (at least if the risk is great), I may be morally required to miss an important appointment to save an innocent life (but not to prevent a stranger from suffering a very minor and transient harm). We can summarize by saying that the general obligation to prevent harm is limited to efforts that incur reasonable

costs and that what constitutes reasonable costs is determined, in part, by a principle of proportionality.

Individuals have *special* as well as general obligations to prevent harm. Special obligations arise from promises, contracts, and certain relationships. The special obligations to prevent harm among family members may be more weighty than general obligations. This is particularly evident in the case of parents' obligations toward their minor children because it is parents' choices that result in children being brought into the world in a condition of dependency and vulnerability.

Similarly, the duty to prevent harm to one's spouse is generally an especially weighty one because marriage vows pledge mutual support and unique loyalties. Whether an individual has especially weighty obligations to prevent harm to siblings, simply because they are siblings, is less evident. For in this case there is no element of choice as there is in the case of children or spouses.

To extend the "higher cost" principle beyond the relationship between parents and children and between spouses to biological relationships generally is more problematic. The standpoint of our responsibilities to prevent harm is not whether someone is biologically related, or how close the biological tie is, but the character of the interactions with that individual — the legitimate expectations and patterns of reliance, and the debts of gratitude that have arisen from the interactions. Thus, acting ethically may require that I bear a greater cost to prevent harm to my friend, to whom I have no biological tie, than to my brother, from whom I have been separated since early childhood.

In some cultures, relationships among relatives beyond the nuclear family are closer than in other cultures. Members of the extended family are connected by a dense web of on-going cooperative interactions. These patterns of cooperation can generate special obligations that would not otherwise exist, and they can add weight to the special obligations that typically exist in all families. The special obligations characteristic of close extended families have their source both in the need to avoid thwarting the legitimate expectations of mutual aid which these on-going patterns of cooperation generate and in what is sometimes called justice as reciprocity, according to which

those who accept the benefits of a cooperative enterprise are obligated to do their fair share in sustaining it.²⁸

The extent and robustness of special obligations within families vary not only across cultures, but within cultures. In some families, for an individual to fail to inform the members of his extended family that he has tested positive for a serious but effectively treatable genetic disorder might be acceptable; in others, not informing them would be inconsistent with legitimate expectations of mutual protection. This makes it even more difficult to make sound generalizations about the extent and weight of an individual's obligation to tell others of the results of a genetic test.

Considering the distinction between general and special obligations to prevent harm, as well as the "reasonable costs" limitation and the proportionality constraint on the latter, how does this all apply to the case of "telling" the results of a positive genetic test? Under what circumstances, if any, does a person who has just learned that he has tested positive for a deleterious genetic condition have an ethical obligation to inform biological relations that they are at risk?

Consider a case where John tests positive for the C282Y mutation²⁹ for hereditary hemochromatosis. He knows that he has two copies of the mutation which accounts for the majority of cases of non-acquired iron overload disease. He knows that the disorder is autosomal recessive and understands that this means that his siblings each have a one in four chance of having two copies of the mutation. Assume that he also knows that those who have two copies have a high probability of serious damage if not diagnosed and treated before stored iron levels reach toxic levels. To simplify the case further, assume that the siblings are both males.³⁰ Recall that the treatment for this condition, regular phlebotomies to reduce iron levels, is safe, relatively noninvasive, cheap, and fully effective if undertaken early enough. Finally, because genetic as well as metabolic testing for this disease is, at present, not part of standard medical examinations, we

28. For a discussion of how participation in mutually beneficial cooperative schemes can generate duties of reciprocity according to a principle of fairness, see JOHN RAWLS, *A THEORY OF JUSTICE* 111-12 (1971).

29. The C282Y mutation is the allele that accounts for the majority of cases of hereditary hemochromatosis. See Wylie Burke et al., *Hereditary Hemochromatosis: Gene Discovery and Its Implications for Population-Based Screening*, 9-10 (March 3, 1997) (reporting consensus of a panel of experts convened by the Center for Disease Control and Prevention and the National Institutes of Health, an unpublished manuscript available from National Center for Chronic Disease Prevention and Health Promotion).

30. Females have a natural "de-ironing" mechanism, menstruation. See *id.* at 8. This may account for the fact that women who have hereditary hemochromatosis tend to have less severe symptoms than men. See *id.*

can assume that there is a high likelihood that unless John notifies his siblings that they are at risk, they will not be tested. Therefore, if John's siblings have the disease, it will go undiagnosed until serious and irreversible damage has occurred.³¹

In such a case, the harm to be prevented is great. Those who are informed that they are at risk must act to prevent the harm especially if it is unlikely that the harm will be prevented in any other way. The regimen of phlebotomies itself generally produces no harms of comparable weight to the harm being prevented. At this point, a familiar distinction between *prima facie* obligations and general obligations is clearly relevant. John has a *prima facie* duty, with no commitment to excessively onerous requirements, to warn in all cases where there is a reasonable expectation that serious harm will be prevented. Therefore, John has a *prima facie* obligation to inform his siblings that he has tested positive. Whether he has an obligation, all things considered, to inform them will depend upon whether informing them would impose "unreasonable" costs on John.

The possible risks to John are highly speculative at best. He *may* be stigmatized by other family members for being "genetically defective." A very small possibility exists that whatever risk of insurance discrimination he may have incurred by having tested positive *may* be increased if his siblings are tested and one or more test positive and if, somehow, a data bank of medical records identifies John as being at risk because he is related to someone who is known to have tested positive.

It is worth noting that the evidence concerning insurance discrimination against those diagnosed with hereditary hemochromatosis is mixed. There are self-reporting cases³² of discrimination.³³ However, there is also anecdotal evidence that some individuals undergoing treatment have succeeded in convincing insurers to overturn initial decisions to withhold coverage.³⁴ Most importantly, many states now have laws prohibiting or limiting insurance discrimination on ge-

31. Hereditary hemochromatosis is frequently not diagnosed until damage is irreversible, and neither the metabolic (serum ferritin or transferrin saturation) test for it or the newer genetic (polymerase chain reaction) test are at this point widely employed. For a valuable overview of the epidemiology, diagnosis, and clinical management of hereditary hemochromatosis, see generally, HEMOCHROMATOSIS (James C. Barton & Corwin Q. Edwards eds. 1988); see also Burke et al., *supra* note 29, at 5-6.

32. Self-reporting refers to surveys in which individuals are asked whether they believe they have been subject to genetic discrimination.

33. See Billings et al., *supra* note 2, at 478.

34. Interview with James C. Barton, Director, Southern Iron Disorders Center, Birmingham, Alabama (Aug. 1997).

netic grounds.³⁵ Furthermore, the Clinton administration has called for federal legislation that would prohibit exclusion from insurance on the basis of a positive test for hereditary hemochromatosis or any other genetic condition.³⁶

Available data on the frequency of genetic stigmatization and its psychological effects on those stigmatized is if anything even less satisfactory than data on genetic discrimination in insurance. It seems reasonable to expect, however, that some genetic conditions are more apt to stigmatize than others — genetically-based Alzheimer's dementia or genetically-based breast cancer may stigmatize quite differently than excessive iron storage. Some anecdotal evidence indicates that hereditary hemochromatosis patients do not tend to experience stigmatization.³⁷ Furthermore, it cannot be assumed that whatever stigmatization hemochromatosis patients may experience is so serious as to exceed the "reasonable cost" proviso on the obligation to prevent harm. A reasonable cost to bear is proportional, within limits, to the seriousness of the harm to be prevented, and the harm to be prevented in the case of hereditary hemochromatosis is very grave.

There is another possible cost to John. If his relationship with one or both of his siblings is strained or in conflict, then it may be very difficult for him to communicate the information effectively.³⁸ It is important to emphasize that John cannot be expected to *successfully* convey the information (anymore than a physician can be expected to successfully make a patient understand the risks of a procedure). John's obligation is to make a good faith effort to provide his siblings with information which may enable them to avert a grave harm.

In some cases, discharging this obligation may be relatively easy. The best way for John to discharge his obligation to inform may simply be to tell his siblings that he has tested positive which means that they are at a higher than average risk for the disease. In other cases, if John has good reason to believe that there is some question as to whether they would want to be informed, it may be more appropriate for him to tell them that he has received some information about his own health status that is relevant to their own and leave it up to them

35. See Rothenberg, *supra* note 22, at 313-17 (describing fifteen state statutes and the extent to which discrimination is prohibited).

36. See The White House Briefing Room (visited Jan. 20, 1998) <<http://www.whitehouse.gov/WH/html/briefroom.html>>.

37. Interview with Peter Beatty, M.D., Meriter Hospital, Oncology and Hematology Department, Madison, Wisconsin (Feb. 1996); interview with James C. Barton, Director, Southern Iron Disorders Center, Birmingham, Alabama (Aug. 1997).

38. Personal oral communication with Gail Geller, Associate Professor, Johns Hopkins Medical Institution and Institute of Biomedical Ethics (Aug. 1995).

to decide whether to inquire about the specific character of the information.

Given the facts of the case as described, we may conclude that John has an obligation to inform his siblings that he has tested positive, unless he has some special reason, based on his knowledge of his siblings' preferences, to take a more guarded approach. He has this obligation regardless of whether he has a "special" relationship with his siblings (as opposed to his children or spouse). John is in a position to prevent serious harm because he is a sibling of the other individuals. However, he would still be obligated to inform his siblings of his positive test results if he had been separated from them at birth, and had no relationship with them but somehow knew how to contact them with the news of his test. What is crucial for his responsibilities is the fact that he can act to prevent serious harm which is not likely to be prevented otherwise and can do so with little cost to himself.

The conclusion that John should inform his siblings of the results of his test assumes, of course, that they have not instructed him that they do not wish to be informed and that he has no other good reason to believe that they do not wish to be informed. The obligation to prevent harm is limited by respect for the autonomy of competent individuals. In the case of some genetic tests, such as the test for Huntington's disease,³⁹ some people do not wish to be informed that they are at risk.⁴⁰ Given the dreadfulness of the condition and its incurability, this preference not to be informed of the risk is understandable. The importance of not overgeneralizing across ethically different tests is clear.

Earlier I noted that one ethically relevant institutional fact is the risk of insurance discrimination and that this fact may change if the use of genetic information for exclusion is effectively prohibited.⁴¹ The preceding example includes another institutional fact that shapes our conclusions about John's responsibilities: The current health care system is not doing a good job of diagnosing hereditary hemochromatosis or of making people aware of the need to be

39. Huntington's disease is a "rare hereditary disease characterized by quick involuntary movements, speech disturbances and mental deterioration." BENJAMIN F. MILLER & CLAIRE B. KEANE, *ENCYCLOPEDIA & DICTIONARY OF MEDICINE, NURSING, & ALLIED HEALTH* 706 (5th ed. 1992). Huntington's disease appears in adults, ages 30 to 45, causes rapid deterioration, and usually results in total incapacitation or death. *See id.* at 706-07. No cure exists and patients usually need to be institutionalized. *See id.*

40. *See* Ann-Marie Codori et al., *Self-Selection in Predictive Testing for Huntington's Disease*, 54 *AM. J. MED. GENETICS* 167, 170 (1994).

41. *See* Billings et al., *supra* note 2, at 481 and accompanying text.

tested.⁴² If John's siblings and all other males over the age of forty were tested routinely for hereditary hemochromatosis as part of a program of periodic physical exams made available by a national health care system, one important fact in our scenario would change: It would not be the case that the harm John seeks to prevent would likely occur unless he acts to prevent it.

John's ethical responsibilities seem straightforward: He ought to inform at least his siblings and to suggest that they inform their children, especially if they are male, that they are at much greater than average risk for liver and heart disease. Matters get more nebulous as John begins to consider how far his obligation to inform relatives extends. Should he notify the offspring of his deceased father's deceased brother? If John is homozygous⁴³ for the mutation, then the probability that everyone in his extended family is at greater risk than the general population is increased. Does this mean that he has an obligation to inform all of them? What if doing so involves considerable time and expense?

No determinate answer to these questions exists. The boundaries of the obligation to prevent harm, whether we consider it a general obligation or a special one, are not clearly demarcated. However, the more remote the biological ties, the less likely that the relative will have the medical condition. In addition, the more remote the relative, the greater the cost for John to locate and notify those relatives. In general, as the circle of biological relatives expands, John's obligations attenuate. It is not possible to locate a precise point at which he no longer has an obligation to make any effort to inform.

There is another reason to conclude that John is not obligated to inform the most distant relatives of the results of his test: He may, in fact, be obligated *not* to contact them in consideration of their privacy. The lower the probability that they will be affected, the more the value of their privacy weighs against informing them.

Nevertheless, a significant obligation exists in some circumstances to inform others of the positive results of a genetic test (e.g., the hereditary hemochromatosis case described above). The principle moral obligation to inform others of the results of one's positive genetic test is the obligation to prevent harm. It is important to determine whether one actually has an obligation (as opposed to a *prima*

42. See Burke et al., *supra* note 29, at 7.

43. In this context, "homozygous" means having two copies of the mutated gene. See CHURCHILL'S MEDICAL DICTIONARY 289 (2d ed. 1989).

facie obligation). If an obligation is present, then the extent of the obligation depends upon the following factors:

1. The severity of the harm to be prevented (the more severe the harm, the weightier the obligation);
2. The probability that the harm will occur if the individual is *not* informed (the higher the probability, the weightier the obligation);
3. Whether there is a "special" relationship between the individual who is in a position to inform and the one who would benefit from being informed (Special obligations generally are weightier in the sense that they count as "reasonable" higher costs.);
4. The costs to the individual who will be informed of preventing the harm (e.g., a regimen of phlebotomies in the case of treatment for hereditary hemochromatosis versus preventive surgery in the case of colorectal cancer or perhaps preventive mastectomy in the case of BRCA1) (the lower these costs, the greater the net benefit of being informed and the weightier the obligation to inform); and
5. The costs of informing to the informer, including primarily: (i) the risk of insurance or employment discrimination (the probability that exclusion from coverage or loss of job opportunities will occur times the cost of exclusion or loss of job opportunities to the individual excluded), and (ii) the risk of stigma (the probability that the one who informs others of his positive test will be stigmatized times the magnitude of the harm to him of being stigmatized) (the lower these costs, the stronger the case for saying that there is an obligation to inform).

B. Less Favorable Cases for Attributing an Obligation to Inform Relatives

Consider a quite different situation, at the other end of the spectrum of responsibility, where there is no clear obligation to inform relatives at risk. Suppose that Mary is tested for the APO E4 "Alzheimer's gene" and is told that she is homozygous for it. She is also told that there is at least a 90 percent probability that she will have Alzheimer's disease by the age of 65 (and an even higher probability if there is a significant history of this disease in her family). Is Mary obligated to tell her relatives of the results of her test?

Assume that the obligation to prevent harm is the most significant source of a possible obligation to tell. Since no effective treatment for Alzheimer's is currently available and learning that one is at much higher than average risk may be very distressing, then the case

for informing relatives is very weak. Furthermore, the cost to Mary of informing others is likely to be higher than John's cost of informing others. The stigma of Alzheimer's may be greater because it is a disease that destroys the characteristics of persons that we value most. In addition, the risk of insurance discrimination, especially for long-term care insurance, may be considerably greater.

This is not to say that there are no benefits of being tested for the "Alzheimer's gene," nor that there are no benefits that Mary's relatives might derive from knowing about the positive results of her test. Some physicians and spokespersons for Alzheimer's organizations have wrongly assumed that if there is no medical benefit to be derived from testing (because there is no effective therapy), then there is no benefit.⁴⁴ This assumption is mistaken for two reasons. First, some who test for the "Alzheimer's gene" will test negative, and a negative result may relieve anxiety. Second and perhaps more importantly, for some individuals, knowing that they have a very high probability of becoming severely disabled and cognitively impaired at a relatively early age may be a benefit, despite the psychological distress it causes, because this knowledge will lead them to plan their lives differently. For example, if one knows that there is a very high probability that one will have Alzheimer's by age sixty-five, one may have children at an earlier age, in order to be able to enjoy more years with them and with grandchildren, one may choose shorter term investments, retire earlier, or even choose a different career.

Those who overlook these benefits and focus only on the psychological costs of a positive test and the fact that there is now no effective treatment, commit a classic fallacy that is characteristic of medical paternalist thinking:⁴⁵ They reduce benefits to medical benefits, or they wrongly assume that what they themselves would not regard as beneficial cannot be beneficial to the patient.

When Mary contemplates whether to inform her relatives of her positive test, she cannot assume that simply because there is no effective treatment, there is no benefit that her relatives could derive from knowing her test results. Some of her relatives might wish to know, and if they knew, they might take the test themselves. Nevertheless, even if Mary would like to know whether she has the gene because she

44. Personal oral communication with Tikka Beard, spokesperson for the Utah Alzheimer's Organization (Apr. 1996).

45. Here medical paternalist thinking should be understood to mean thinking that assumes that physicians may interfere with the liberty of action or freedom of information of competent patients for the patients' own good. See Allen Buchanan, *Medical Paternalism*, 7 PHIL. & PUB. AFFAIRS 370, 372 (1977).

wants to be able to plan her life differently if she does, she cannot assume that others will feel the same way. It is a different matter if she knows that some particular relative to whom she is very close definitely would want to know if he is at higher than average risk. She can assume, however, that most people will be distressed to learn that a relative, especially a close relative, has the "Alzheimer's gene" and that consequently they are at risk for this terrible, incurable illness. Given this constellation of factors, it would be implausible to argue that Mary has an obligation to tell even her close relatives about the results of her test.

Genetic testing for hereditary hemochromatosis and for APO E4, or for Huntington's disease lie at the opposite extremes of a spectrum of responsibility. Other tests, including some that are likely to become available in the next decade, present a much more ambiguous picture for the ethics of telling and testing. Where the prospects for effective treatment are not so certain as with hereditary hemochromatosis nor so bleak as with Alzheimer's, or where there the risk of stigma or of insurance or employment discrimination is neither negligible nor great, sorting out whether, or to whom, one has the obligation to tell may be a very complex matter. Where the balance of reasons lies in any particular case may also depend upon whether genetic testing has become standard medical practice and upon whether those who might benefit from being told have access to genetic testing. If the tests in question are part of standard medical practice and if everyone has access to them, then there may be no duty to warn relatives at risk because they will have the opportunity to be tested themselves if they wish.

It is likely in the future that the number of genetic tests for genetic susceptibilities to disease will increase. In many cases there will be no single, fully effective treatment for the condition. Instead, a positive test for the susceptibility will alert the patient to prepare a complex strategy of special medical surveillance for early detection of symptoms, dietary and life style changes, and possibly pharmaceutical interventions. In such cases the causal chain between receiving information that one is at risk and preventing the harm is more complex. Some of the crucial links in the chain depend upon choices the patient makes over time (such as whether to change his or her lifestyle to reduce risk). For any number of reasons, including disruption of medical coverage due to unemployment or lack of self-discipline, the patient may not carry out the needed life style modifications or continue the medical surveillance. Consequently, a person who has been diagnosed with a genetic susceptibility for which the reduction of risk

depends upon this complex array of factors may be much less confident that harm will be prevented if he informs his relatives at risk of the results of his test.

IV. CLINICIANS' ETHICAL RESPONSIBILITIES

A. *Obligations Regarding the Decision to Test Versus Obligations Once the Test Results are Available*

As more and more genetic tests become available, clinicians will be faced repeatedly with the question of whether they should recommend a particular test to a particular patient or whether they should even inform the patient of the existence of the test. Clinicians are obligated to recommend tests or notify patients of their availability if doing so is required by sound standards of medical practice.⁴⁶

Relying upon standards of medical practice to determine the physician's obligation is not sufficient, however, in an area such as genetic testing, where no coherent standards of practice exist that are capable of accommodating the flood of new tests that will become available in the coming years. When practice standards are unclear or rapidly changing, clinicians must rely upon their judgment about what would best serve the patient's interests, within the constraints of respect for the patient's autonomy and privacy.

In addition, whatever standards of practice eventually evolve are likely to provide only very general guidance at best because, as our analysis has already shown, it is difficult to generalize across types of genetic tests. For the fortunate cases where the probability of serious pathology and of safe and effective treatment are both high, sound medical care will require that patients at high risk for the condition be offered the option of testing. Even in these relatively unproblematic cases, there will be disputes about what constitutes high risk. In any system that faces the need to limit medical expenditures, what constitutes high risk must be determined in part by cost, or rather by the ratio of costs to benefits.

In other cases, where the probability of serious pathology or effective treatment is lower or subject to greater uncertainty, it will be much more difficult to forge consensus on a standard of care. It is important to emphasize, however, that there is nothing here that is peculiar to *genetic* testing. Many nongenetic diagnostic tests are char-

46. See John H. Derrick, Annotation, *Medical Malpractice: Liability for Failure of Physicians to Inform Patient of Alternative Modes of Diagnosis or Treatment*, 38 A.L.R.4th 900 (1998).

acterized by these same complexities and ambiguities.⁴⁷ As far as the clinician's obligations are concerned, the fact that positive genetic tests only reveal a probability of future illness does not set them apart from other tests.

B. Old Problems of Paternalism, New Applications

Familiar patterns of medical paternalist thinking can distort the clinician's judgment about whether to offer a test, whether to comply with a patient's request for a test, or whether to inform a patient about the availability of a test. In each of these decisions, clinicians must avoid the error of reducing patient benefit to medical (i.e., therapeutic) benefit and must fix their attention on what is in the patient's best interest, not what would be in the clinician's best interest.

In other words, the clinician is obligated to take the competent patient's view of his own best interests seriously. The clinician must endeavor to know what the patient's values and wishes are. With one exception, there is nothing here that is in any way peculiar to decisions concerning genetic tests. The exception is that the process of informed consent to genetic testing should include the clinician's disclosure that there may be a risk of insurance or employment discrimination. Although this is not a "medical" risk in the same way in which a bladder infection is a risk of cystoscopy,⁴⁸ it is nonetheless something a reasonable decision maker would want to be apprised of.⁴⁹ Given the fact that many patients may be unaware of the risk of insurance discrimination, the person best placed to inform them is the physician.

C. Medical Needs Versus Market-Generated Preferences

If profit-seeking marketers of tests are successful in generating patient demand, clinicians will increasingly see patients who request particular tests or who at least are the first to raise the question of whether they should be tested. Again, this is nothing peculiar to genetic testing. For many years physicians have been faced with patients who request or demand antibiotics for the treatment of the common

47. This is evidenced by the common practice of gaining a second opinion from a specialist.

48. Cystoscopy is a "visual examination of the interior of the urinary bladder by means of a cystoscope." MELLONI'S ILLUSTRATED MEDICAL DICTIONARY 120 (2d ed. 1985).

49. "A risk is thus material when a reasonable person, in what the physician knows or should know to be the patient's position, would be likely to attach significance to the risk or cluster of risks in deciding whether or not to forego the proposed therapy." *Canterbury v. Spence*, 464 F.2d 772, 787 (D.C. Cir. 1972).

cold, even though the treatment is inefficacious. More recently, pharmaceutical companies, in magazine and television advertisements, have been appealing directly to individuals to "ask your doctor about____."50 The advertisements for mail-in hemochromatosis tests noted above51 are directed not primarily at physicians, but at consumers. The fact that these issues are familiar in nongenetic contexts does not make it easier to determine the clinician's obligations. Clinicians must steer a course between reverting to the paternalism of an earlier era and becoming passive vendors of services who respond unquestionably to patient demands, or even worse, becoming marketers who stimulate consumer demand for the sake of profit, and thereby forsake the notion of medical need.

To revert to paternalism is to fail to take the competent patient seriously as a partner in decision making and as a person whose own estimate of his best interests ought to be honored. Simply to respond to patient demand, no matter how misguided, or to ignore the distinction between preference and medical need, is to abdicate professional responsibility by refusing to exercise professional judgment.

D. *The Clinician's Obligations After the Patient Tests Positive*

I will assume what I hope is, at this stage in the development of medical ethics, uncontroversial: If a competent patient tests positive, then he or she is to be informed. Instead of re-enacting familiar battles against medical paternalist justifications for not informing patients of test results, I wish to focus on what the responsibilities of the clinician are after the disclosure of a positive result has been made to the patient.

The most obvious questions are these: (1) Should the clinician advise or urge the patient to notify relatives at risk? (2) If the patient refuses to do so, should the clinician notify them?

E. *Nondirectiveness, an Outmoded Norm?*

Answering the first question requires a critical assessment of the norm of nondirectiveness. This norm — which has achieved the status of a dogma — was developed in the peculiar context of reproductive-predictive testing.⁵²

50. See, e.g., Advertisement, *Depression saddens. Prozac can help.* HEALTH, NOV./DEC. 1997, at 20-22; Advertisement, *Zocor*, HEALTH, NOV./DEC. 1997, at 27-28.

51. See *supra* text Section II.A.

52. See Geller et al., *supra* note 7, at 1120 and accompanying text.

It is important to understand that the norm of nondirectiveness is not a fundamental principle of morality, nor is it a norm that is generally followed or thought to be appropriate in any other area of medical practice. In most cases, patients expect their physicians to recommend a course of action in response to a positive test. For example, if a patient is told that she has cancer, she will generally expect that the physician will suggest a course of treatment.

The norm of nondirectiveness is the product of a certain historical experience. Earlier we sketched the broad outlines of its origins.⁵³ Nondirectiveness was nourished by the peculiar context of reproductive decision making, in the midst of the controversy over abortion, and in an effort to dissociate modern genetics from the taint of eugenics.⁵⁴ Once one leaves the context of reproductive-predictive testing, it is much less plausible to advocate nondirectiveness — unless one thinks that physicians should be more concerned about avoiding any hint of an association with eugenics than about helping patients understand that they, like other human beings, have ethical obligations.

Applied to the situation of a positive diagnostic-predictive test, the norm of nondirectiveness imposes limits on what the clinician may communicate to the patient about the nature of the choices confronting the patient. Here, it is useful to distinguish two different understandings of nondirectiveness: strong and weak. According to strong nondirectiveness it would always be wrong for the clinician to tell the patient that the patient has an obligation to inform even her closest relatives of her positive genetic test. According to weak nondirectiveness, it may be appropriate or even obligatory for the clinician to tell the patient that she ought to inform relatives at the highest risk (e.g., siblings), but the physician should not go beyond informing the patient of what the he takes her obligation to be. For example, pressuring the patient to inform by implicitly threatening to break off the therapeutic relationship or by threatening to inform the relatives if she does not, is impermissible according to weak nondirectiveness.

It is probably fair to say that the orthodox doctrine, at least among genetic counselors in this country, is that strong nondirectiveness is the appropriate norm⁵⁵ (whether those who profess it to be

53. See *supra* text Section II.

54. See generally, KEVLES *supra* note 8 and accompanying text.

55. See F.C. Fraser, *Genetic Counseling*, 26 AM. J. HUM. GENETICS 636 (1974) (discouraging communication of anything but the facts in genetic counseling); M. Yarborough et al., *The Role of Beneficence in Clinical Genetics: Nondirective Counseling Reconsidered*, 10 THEORETICAL MED. 139 (1989) (observing that the dominant view of genetic counseling insists that it

always comply with it is another matter). The justification for nondirectiveness is the need to avoid the clinician's "imposing his values" on the patient.⁵⁶ It does not follow, however, that one need to go as far as strong nondirectiveness to avoid "imposing values."

Generally speaking, it would be hyperbole to say that when one individual tells another that he should do something, there is an imposition of values. In fact, there is something odd — indeed something demeaning and disrespectful — about the assumption that merely offering such a judgment about a person's obligations shows disrespect or is an infringement of autonomy. It is to assume that the patient is a very fragile vessel that will be shattered at the very sound of normative language.

Depending upon whether it comes from a physician or a layperson, the judgment that the patient has an obligation to inform relatives at risk that the patient has a genetic disease may have quite different effects. Because of what might be called "status-deference" toward physicians, the physician risks imposing her values if she simply states that the patient has an obligation to inform her relatives.⁵⁷

It is true that in the United States until very recently, physicians *qua* physicians have been treated as figures of considerable authority. However, many patients have begun to take a more critical attitude toward their physician's statements and recommendations.⁵⁸ With some patients, status-deference may increase the risk that a patient will feel pressured into doing what the physician states that the patients should do. Nevertheless, a physician may reduce this risk by being careful to make it clear that the patient will not be penalized if he chooses to ignore the physician's view about what his responsibilities are. To the extent that status-deference toward physicians varies across cultures, the risk that by informing patients of their ethical responsibilities to others, physicians will be imposing their values is culturally-variable as well; the higher the status-deference accorded to clinicians, the higher the risk of clinicians imposing their values.

The primary point is that where the prevention of a serious harm is at stake, one should not assume that clinicians must abstain from communicating to the patient any ethical judgment whatsoever, as if

be done "in a nondirective manner wherein the counselor maintains a morally neutral attitude . . .").

56. See *supra* text Section II.

57. What I call status-deference is to be contrasted with what might be called merit-deference. Status-deference is deference to a physician simply because he or she is a physician. Merit-deference is deference based on a perception of the actual abilities of the individual in question.

58. See PAUL STARR, *THE SOCIAL TRANSFORMATION OF AMERICAN MEDICINE* 17-18 (1982).

reducing the risk of imposing values to zero was the only ethical consideration. Infringing the patient's autonomy by imposing one's values on him is something to be avoided, but it is not to be avoided at all costs. Furthermore, it is worth noting that there is a clear sense in which appealing to the patient's moral sensibility, by pointing out what his obligations are, is a recognition of his autonomy, not a denial of it. As Kant observed, moral imperatives are directed toward moral agents, beings who have a will that can enable them to act contrary to their inclinations.⁵⁹ Finally, and most importantly, much will depend upon how the physician communicates to the patient his belief that the patient has a moral obligation to notify relatives of the test result. There is a great difference between asking questions that stimulate the patient to arrive at the conclusion that she has an ethical obligation, or if this fails, stating in a calm fashion that one believes that she does have an obligation, and subjecting the patient to a moralizing harangue that borders on intimidation or that impugns her character.

The case of John, who tested positive for hereditary hemochromatosis, provides the clearest instance of a situation in which the adherence to strong nondirectiveness is not morally defensible. Suppose, that after explaining the significance of a positive result to John, John's physician asks him if he has any children or siblings. John says that he has two male siblings and a male child. The physician then tells John that there is an issue of ethical responsibility to be considered. John does not respond to this invitation to consider what his responsibilities are. Surely the physician should suggest to John that he should inform his siblings and son that they are at unusually high risk for a disease that is lethal if not treated. The physician should suggest to John that he should disclose his test results especially since there is a safe and effective treatment, and because, at present, many cases of this disease go undiagnosed until serious, irreversible damage has been done.

As suggested earlier, there is another way one can argue against strong nondirectiveness. One might try to marshal survey data to attempt to show that, in fact, most patients expect their physicians to exceed the strictures of strong nondirectiveness. They expect and desire their physician to be a moral counselor while showing respect and not exerting coercive pressure. There is, however, an independent case for abandoning the nondirectiveness norm: There are instances in which a patient's ethical obligation to inform relatives is clear and

59. See Immanuel Kant, *Groundwork of the Metaphysic of Morals* 62-64 (H.J. Paton trans. 1948) (1792).

uncontroverted, in the light of the widely accepted principle that we ought to prevent serious harm. Informing the patient that this is so need not involve "imposing values."

In some instances, as is presently the case with the tests for Huntington's, Alzheimer's, or BRCA1 or BRCA2, there will be no clear obligation to inform, and it would be presumptuous and unwarranted for the clinician to tell the patient that he should inform anyone of the results of his test. In rejecting strong nondirectiveness, I am not saying that in every case, or even in most cases, the clinician ought to tell the patient what his obligations are, much less that the clinician should try to impose a sense of duty on the patient.

Everything will depend upon the particular constellation of the factors noted above. Instances in which the strongest case can be made for attributing an obligation to the patient to inform others at risk will, in general, be those in which it will also be appropriate for the clinician to tell the patient that he does have such an obligation.

So far, I have argued against strong nondirectiveness. A rejection of strong nondirectiveness does not entail that the clinician should inform her patient's relatives that they are at risk if the patient refuses to do so. John is the physician's patient, not John's relatives. John's physician has no special obligation to inform John's relatives.⁶⁰ Moreover, John's physician has a duty of confidentiality with respect to John, by virtue of his role as John's physician.⁶¹

Whether, despite this obligation of confidentiality, it would be *permissible* for John's physician to inform John's relatives when John refuses to do so is a more difficult question. On the one hand, to inform John's relatives of his test result without his consent would be to breach the physician's *role-specific obligation* of confidentiality. On the other hand, as we have already seen, human beings have general obligations to prevent harm, and this seems to imply that John's physician should inform John's relatives, assuming that John refuses to, if doing so is likely to prevent serious harm.

The strongest argument for saying that it would be impermissible for John's physician to inform John's relatives without John's consent

60. *But see* Safer v. Pack, 677 A.2d 1188 (N.J. Super. Ct. App. Div. 1996) (holding that a physician has a duty to warn those known to be at risk of avoidable harm from a genetically transmissible condition).

61. A physician's legal duty to maintain confidentiality arises from licensing statutes, case law, and medical records statutes. *See, e.g.*, Humphers v. First Interstate Bank of Oregon, 696 P.2d 527 (Or. 1985); Felis v. Greenberg, 51 Misc.2d 441 (N.Y. Sup. Ct. 1966); Berry v. Moench, 331 P.2d 814 (Utah 1958). *But see*, Moses v. McWilliams, 549 A.2d 950 (Pa. Super. Ct. 1988) (holding that a patient's right to confidentiality is not absolute, e.g., patient's personal injury action against the doctor).

is that a proper understanding of the role of physician includes a special fiduciary obligation to his patient that extinguishes, or, at least significantly weakens, what would otherwise be a general obligation to prevent harm to innocent persons. On this view, the physician's special, role-specific obligation of confidentiality toward his patient outweighs his general obligation to prevent harm to others.

Even if it is generally true that the role-specific obligation of confidentiality takes precedence over the general obligation to prevent harm to third parties, it would be implausible to hold that this, or any role-specific obligation is absolute. The role-specific obligations of physicians are elements of a particular social practice. The ethical justification for a particular role-specific obligation, such as the physician's obligation to preserve confidentiality regarding medical information about his patient, is justified ultimately by demonstrating that the social practice in which that obligation is an element promotes human welfare and freedom and does so without unacceptable infringements of important rights.

Yet one may acknowledge the ethical value of a social practice in which physicians generally give precedence to special obligations of confidentiality toward their patients and still concede that there can be individual cases in which the physician's acting so as to prevent harm to third parties would be morally preferable. In the well-known *Tarasoff*⁶² case, legal recognition was given to this belief that role-specific obligations of confidentiality are not absolute — they can be overridden in the name of preventing grave harm to a third party.⁶³ It is one thing to say that one can describe situations that constitute legitimate exceptions to a generally appropriate role-specific rule of giving precedence to obligations of confidentiality; it is quite another to assume that those occupying the relevant role (in this case physicians) can generally be relied upon to judge accurately and impartially whether the "exception conditions" are satisfied. In some cases, legitimate exceptions to generally acceptable rules can be specified and built into the rules as exception clauses. In other cases, an exhaustive specification of exceptions would either be impossible or would undermine the effectiveness of reliance on the rule by making it too complicated to apply or by providing too many opportunities for error or abuse in its application. In the latter types of cases, the best is that the individuals who occupy the relevant roles will steer a course be-

62. *Tarasoff v. Regents of the University of California*, 551 P.2d 334 (Cal. 1976) (holding that a psychiatrist had a duty of care to protect the third party against whom his patient had threatened bodily harm).

63. *See id.*

tween slavish adherence to general rules and a hubristic tendency to over-estimate their own ability to identify appropriate exceptions.

Once again, noting the broad spectrum of genetic testing situations proves illuminating and cautions against unsatisfying generalities. In circumstances where there is a very high probability that informing a relative will avert a great harm, where it is very unlikely that the harm will be averted if the individual is not informed, and where the risk to the individual whose confidentiality is breached is small, a physician may be justified in overriding his role-specific obligation of confidentiality. This is only to say that in this exceptional case, it would be morally permissible for the physician to do so, not that breaching the confidence would be ethically required.

The only case of which I am aware that may fit this description is that of hereditary hemochromatosis.⁶⁴ However, reflections on the ethical essentials in the case of PKU testing may also lend support to the claim that the physician's obligation of confidentiality is not absolute. In PKU testing, it is the individual infant who is tested for the disorder.⁶⁵ Moreover, many states perform testing and treatment without the consent of the infant's parent or guardian.⁶⁶ Suppose, however, that the only way to detect this metabolic disorder in the child was to test the mother, but that all the other facts were the same — the disorder has devastating consequences if not treated, the treatment is effective, and if the test is not done, then it is very unlikely that the harm caused by the disorder will be avoided. If this was the case, surely one would not regard the mother's right to confidentiality as absolute. If she refused to authorize transmitting the test results to the State Department of Public Health or some other agency charged with seeing that the treatment is undertaken, one would think it justifiable to inform them anyway.

At present, this is not the way genetic tests work. Tests are either performed on the individual in whom the disorder may be detected, and the prospect that harm will be prevented by informing relatives of the results is uncertain; or the tests are performed on prospective parents to detect risk in offspring, but the only way to prevent the harm is to prevent the child from being conceived or born. It is conceivable, however, that this will change. Prenatal genetic tests might be devel-

64. See *supra* note 4 and accompanying text.

65. PKU (phenylketonuria) is a metabolic disorder that can result in mental retardation. See DORLAND'S MEDICAL DICTIONARY 1278 (28th ed. 1994). Damage can be avoided by a special diet.

66. See Ellen Wright Clayton, *Screening and Treatment of Newborns*, 29 HOUSTON L. REV. 85, 98-99 (1992).

oped which would predict a very high probability of serious but wholly avoidable harm. If such prenatal tests are developed, it will be very difficult to defend an absolute prohibition on breaches of physician-patient confidentiality.

However, it bears emphasis that the situation is entirely different in most actual cases of genetic testing, at this point in the development of the technology. In virtually every existing case, with the possible exception of testing for hereditary hemochromatosis, overriding the role-specific obligation of confidentiality would not be justified, either because the harm to be prevented is not highly probable, or because those who are vulnerable to it are likely to learn that they are at risk in some other way, or because there is no way to avoid the harm even if the person is informed that she is at risk. Accordingly, it would not be plausible to assert that a clinician ought to (or even may) inform the relatives of Mary, who has tested positive for the "Alzheimer's gene" (or for BRCA1, or BRCA2, or Huntington's) without Mary's consent. Whether Mary herself should inform relatives that they are at higher than average risk is another matter, and depends upon the factors discussed.⁶⁷

V. CONCLUSION

This essay has explored the ethical responsibilities of patients and clinicians regarding disclosure of genetic test results. I have argued that the obligation to prevent harm grounds ethical responsibilities for both patients and clinicians. Whether such obligations exist in a particular case will depend upon a number of factors which may or may not be present depending upon the particular facts about the genetic disorder for which one is testing. Once this more particularistic or contextualized approach is taken, it becomes clear that the traditional norm of nondirectiveness is not appropriate for all genetic test situations.

67. See *supra* text Section III.