

Disowning Knowledge: Issues in Genetic Testing

Last fall in Chicago, at a conference sponsored by the Alzheimer's Association and the National Institute on Aging, doctors and researchers met to discuss an ethical dilemma that has grown increasingly familiar as advances in diagnostic techniques outstrip the therapeutic abilities of the medical profession. The meeting focused on the use of a medical test for a particular heart condition—a test that can also, in some cases, predict with 90 percent accuracy whether someone will develop Alzheimer's disease by the age of 80. Should patients tested for the heart condition be told of their risk of contracting Alzheimer's disease, when there is little if anything medicine at present can do to prevent or ameliorate the condition?

Some people, including many of those attending the meeting, believe that the answer to this question is no: if the information is of little therapeutic value, it's of little value to the patient as well. It is wrong to burden the patient with troubling news when there is little or nothing that the physician can do about it.

At this stage in the history of medical practice, we may well be surprised to encounter such a response. Over the past few decades there has been an intense

of health professionals deciding whether a patient should know his or her medical condition is now routinely criticized in bioethics courses. Nonetheless, the advent of genetic testing appears to have provoked a resurgence of paternalistic thinking, especially in those cases where doctors can detect the genetic condition associated with a particular disease but are as yet unable to prevent or treat that disease.

The association between a genetic condition and a disease, and so the type of information a genetic test reveals, is subject to considerable variation. With results from the test for a specific mutation at the tip of chromosome 4, we can predict with near certainty whether an individual will suffer from Huntington's disease, a severe late-onset neurological disorder, but we can't yet tell when the disease will occur. With information from the test for mutations of the BRCA1 gene, we can, in particular situations, conclude that an individual has a susceptibility to a specific type of breast cancer, but we don't yet know what other conditions must be in place to trigger this susceptibility. With information from the test discussed in Chicago—a test that detects the presence of the apolipoprotein E genotype—we can, in particular situations, conclude that an individual is at an increased risk of contracting Alzheimer's disease, but there is still some controversy about the relative importance of this risk factor.

Recent concern has largely focused on these last two tests. At the Chicago meeting, the issue was the disclosure of certain additional information from a test already administered. In other cases, professional organizations, as well as some advocacy groups, have proposed limits on the very availability of certain genetic tests. It is argued that tests for certain conditions should be restricted to research settings for the time being and not offered routinely or to all.

Are these proposals based on medical paternalism? Or can restrictions on genetic testing be defended on other grounds? I wish to examine possible justifications for limiting testing, distinguishing between those that are paternalistic and those that are not. I shall then consider the reasons and responsibilities that might influence patients in deciding whether to be tested or to receive genetic information.

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effort to articulate and defend a person's right to be informed of his or her medical condition. Not so long ago, this right was not widely acknowledged. Health professionals generally assumed that, in the case of certain diseases, patients didn't really want to know. Moreover, even if they did want to, they wouldn't really understand the diagnosis; and even if they did want to know and could understand, they would be so psychologically harmed by the information that the result would likely be, if not suicide, then a clinical depression that would interfere with any sort of available care. Over the years the arguments attempting to defend this medical paternalism have been carefully examined and successfully undermined. The very idea

Grounds for Restrictions

A discussion of reasons for restricting genetic testing should begin by acknowledging that there is no *right* to genetic testing. A right to be informed of test results (assuming that such a right exists) would not entail a right to *be* tested. And a "right to health care" (in the usual ways that phrase is understood) is not taken to include a right to have every diagnostic test, including genetic tests, performed. But though there is no right

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to genetic testing, a decision to withhold or restrict certain tests should be based on good public reasons (as opposed to private, economic reasons). This is especially true in the case of genetic tests, since in many cases genetic testing facilities, e.g., those connected

with teaching hospitals, are supported, directly or indirectly, with public funds.

Reasons for restricting certain kinds of genetic tests can be divided into two broad categories. One set of reasons focuses on the *time and resources* that would be lost by the inappropriate use of genetic testing. Given the current state of knowledge, the results obtained from certain tests may include such a high number of false-positives or false-negatives, or be so difficult to interpret, that performing these tests would be a waste of the health professional's or laboratory's time, diverting resources from tests that are diagnostically more useful. For example, research has revealed a large number of possible mutations in BRCA1. Unless a woman's family history implicates a particular mutation in the occurrence of breast cancer, there is no point in testing her for that mutation; whatever the test result may be, it will not be interpretable. Thus, a decision not to offer BRCA1 testing to all women would be defensible on the grounds that widespread testing would needlessly draw upon society's limited resources of expertise and technology. Where the best available evidence shows that a given procedure



*"Son, now that you've turned sixty there are a few
things you should know about the family."*

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would yield no meaningful information, it is entirely appropriate, so the argument goes, to restrict that procedure.

The second set of grounds for restricting the availability of genetic tests focuses on claims about the *social or psychological harms* that individuals might suffer from knowing their test results, where these harms are not offset by any corresponding medical benefit. Indeed, in many cases these harms are considered to be so palpable and the medical benefits so clearly nonexistent that it is assumed people would not want to know their genetic condition even if they had the opportunity.

One widely cited harm of knowing one's genetic condition arises from the prospect of discrimination in employment or insurance coverage. Someone with a known genetic condition indicating a susceptibility to breast cancer might be denied a job or a promotion, or denied health or life insurance, because she is regarded as a health risk and therefore as too great an economic risk. This concern about discrimination chiefly provides a reason why *third parties* should not be given

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access to an individual's genetic information. Yet an individual may well decide to forgo this information in order to maintain deniability. For example, suppose an insurance contract requires the individual to tell all she knows about her genetic condition, so that discovering that any information was withheld would constitute grounds for dismissing later claims. A person in this situation might well decide to remain ignorant, since she can't be penalized for withholding information she doesn't have.

However, a person can maintain ignorance of her genetic condition only up to a point, since genetic tests are not the only source of information about that condition. Standard family medical histories can sometimes tell a good deal, and claiming ignorance of this history may not be possible. If an individual suffers from Huntington's disease, then his or her children have a 50 percent probability of contracting it as well. If a woman's sister, mother, and aunt suffer from breast cancer, then it is likely that the woman is at greater risk than the general population of contracting breast cancer herself. Furthermore, genetic information is not always bad news. Someone who appears to be at risk for a certain disease because of her family history could discover, and so presumably assure an employer

or insurer, that she is in fact not at risk because her test result was negative. Nevertheless, we should acknowledge that there can be perverse incentives to be ignorant, especially in the absence of appropriate laws

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regarding "genetic discrimination" or regulations regarding insurance and preexisting conditions.

A completely different harm that is associated with genetic information has to do with the psychological burden of knowing. Indeed, one writer refers to such information as "toxic knowledge." Unlike concerns about employment discrimination or insurance, fears about the burden of knowing speak directly to the question of the desirability of self-knowledge. For some people, the discovery that they have a genetic condition that places them at an especially high risk of suffering certain diseases could so depress them that the quality, joy, and purpose of their lives would evaporate. Moreover, even if the results of a genetic test were negative, some people might experience the reaction commonly known as "survivor's guilt," as they contemplate the prospects of their less fortunate siblings or other relatives.

The applicability of this reason will vary from person to person. Some people might be able to handle bad news calmly and move on, while others might become irrevocably incapacitated. We are individuals in how we each deal with the disappointments and tragedies in our lives. Genetic knowledge might be extremely toxic for one individual but less so for another. Presumably, however, if a person does raise this issue in his own case, it probably applies.

Deciding for the Patient

It is this last set of reasons, when invoked to justify limits on the availability of genetic testing, that suggests a resurgent paternalism with respect to medical information. They involve explicit judgments by medical professionals about what would be good for the patient, where the "good" (i.e., the avoidance of certain social and psychological harms) extends beyond matters of medical expertise. Whatever force they may have as reasons an *individual* might give for not wanting to know genetic information, their persuasiveness weakens considerably when they are offered by third parties as reasons for restrictions on genetic testing. While certain people might be psychologically devastated by their test results, there is no evidence to support the assumption that most people will be so devas-

tated; indeed, such an assumption flies in the face of our commonsense knowledge of people's differences. Similarly, the likelihood that people will confront employment discrimination or insurance problems, and the seriousness with which they regard such a prospect, will vary with circumstances. It is therefore paternalistic to cite these concerns as grounds for restricting genetic testing.

The same can be said of arguments that the results of genetic tests are too complex or ambiguous for patients to understand. Test results may identify risk factors rather than yield predictions; the information may consist of probabilities rather than certainties. In other medical contexts, however, the complexity of information is not accepted as an excuse for taking decisions out of the patient's hands. For example, we require physicians to obtain informed consent before they engage in an intervention. However complex the relevant information might be, usefully communicating it to the patient is a challenge to which the professional must rise.

A rejection of the paternalistic arguments does not yield the conclusion that all genetic tests should be available to the public. As we have seen, restrictions on the availability of certain genetic tests, or of any medical procedure, need not be based on paternalism. For example, none of these comments affects the legitimacy or persuasiveness of the scientific reasons for restricting certain tests.

Unfortunately, some of the professional organizations and advocacy groups seeking to restrict genetic testing have allowed an admixture of paternalism to enter into what would otherwise be sound scientific

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arguments. Instead of simply pointing out that a test for BRCA1 mutations can yield no useful information about most women, they express worries about the "fear" and "panic" that widespread testing might provoke. The first objection to indiscriminate testing is valid; the second is not. By including arguments that would in other contexts be rejected as unwarranted medical paternalism, these organizations have inadvertently ceded the moral high ground to the for-profit laboratories that have rushed in to perform these tests. Whether the labs can provide testing with the appropriate care and counseling is an open question. But efforts to regulate or even comment upon their services are likely to be ineffectual so long as the laboratories can self-righteously affirm the patient's "right to know" against the paternalism of their critics.

Similarly, when the researchers in Chicago tried to formulate a policy regarding the disclosure of test results, paternalistic assumptions clouded the issue. It was agreed that a cardiac test yielding information about the risk of Alzheimer's disease poses an ethical problem for the physician, who must either inform patients of their condition or withhold that information. But there is another alternative: the physician can tell patients, before testing for one condition, that information about another condition will be available. Whether or not to be informed becomes the patient's decision. Indeed, this option is standard in communicating the results of various medical tests, including results where disease is not at issue. The obstetrician performing amniocentesis doesn't typically agonize over whether to inform the couple of the fetus's sex. The couple are simply asked whether they want to know. And in our society at this time, the patient's desire to know or not to know is taken to settle the matter.

A Responsibility to Know

It is mainly those who wish to know their genetic condition who are likely to object to paternalistic restrictions on genetic testing. We cannot assume, however, that most people would fall into this category. In

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one recent study, only 43 percent of research subjects who were offered the BRCA1 test agreed to have it performed. Many who refused the test cited the concerns about employment and insurance that I have already described, while others pointed to the psychological distress that knowledge might bring.

If the challenge to medical paternalism is based on the notion that people should be free to make their own choices with respect to information, then in general the decision not to know should be as fully respected as the decision to know. No one would be in favor of frog-marching people to a genetics lab, having

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them tested, and then compelling them to listen to the results. The widely acknowledged right people have to refuse treatment surely includes a right to refuse diagnostic tests. If some people simply don't want their decisions about how they live their lives to depend upon genetic information, it would seem that they have no reason, and certainly no obligation, to know.

Nevertheless, there are many circumstances in which people might have a moral responsibility to know—a responsibility that grows out of their professional or personal obligations. The case for professional obligations, though limited, is fairly clear. The same reasoning that supports drug testing of individuals in particular professions—air traffic controller, train conductor, airline pilot—also supports claiming that these individuals have an obligation to know their genetic information. If an individual might have a condition that, if manifested, would interfere with his job performance in such a way as to endanger other people, that person has an obligation to know and monitor that condition, whether he wants to or not.

Since most of us are not employed in such professions, however, this obligation attaches to relatively few people. Moreover, most genetic conditions are unlikely to have an impact on the safety of other people. It is difficult to argue that an airline pilot's refusal to know whether she is at special risk of contracting breast cancer would endanger the lives of the passengers.

The ways in which personal obligations may generate a responsibility to know one's genetic condition have not been given comparable attention, even though they are more widely applicable. Most of us are enmeshed in a network of personal obligations and commitments—to families, dependents, loved ones. In many cases, with information about our medical condi-

tion, we can more effectively discharge our obligations, or at least avoid measures that, under the circumstances, may be futile. Consider the case of a 50-year-old parent of minor children who refuses to know whether he is at high risk of contracting Alzheimer's disease within the next ten years. His refusal to know might be irresponsible; it might amount to a failure to engage fully in the (not just financial) planning that is part of a parent's commitment to his children. Whether one has a moral responsibility to know one's genetic condition, and the strength of that responsibility, will depend upon the particulars of the situation. In all likelihood, however, a person's *responsibility* to know will not depend upon the strength of his or her *desire* to know or not to know.

The idea of having a responsibility to know can seem jarring at first. We are drawn to a picture of an individual, faced with the prospect of knowing, weighing how that knowledge would affect her personally. The thought that someone ought to know seems to go against our cultural assumptions, as if such an obligation were an unwelcome interference in the private relationship a person has with her own life. The problem with this picture of solitary individuals contemplating whether to know about their future is that it fits so few of us.

How should the responsibility of knowing be balanced against the possible burden and cost of knowing? There is probably little of use that can be said at this level of generality, since much will depend on the circumstances. The 50-year-old who has minor children, by birth or adoption, is in a different situation from the footloose 20-year-old. In any event it should be clear that if we are to make responsible decisions about accepting or refusing medical information, we must begin by acknowledging that these decisions affect others as well as ourselves.

—Robert Wachbroit

Sources: Gina Kolata, "If Tests Hint Alzheimer's, Should a Patient Be Told?" *New York Times* (Oct. 24, 1995); Rick Weiss, "Tests' Availability Tangles Ethical and Genetic Codes," *Washington Post* (May 26, 1996); American College of Medical Genetics/American Society of Human Genetics Working Group on ApoE and Alzheimer Disease, "Statement on Use of Apolipoprotein E Testing for Alzheimer Disease," *JAMA*, vol. 274, no. 20 (1995); National Advisory Council for Human Genome Research, "Statement on Use of DNA Testing for Presymptomatic Identification of Cancer Risk," *JAMA*, vol. 271, no. 10 (1994); Charles Siebert, "The DNA We've Been Dealt," *New York Times Magazine* (Sept. 17, 1995); Caryn Lerman, et al., "BRCA1 Testing in Families with Hereditary Breast-Ovarian Cancer," *JAMA*, vol. 275, no. 24 (June 26, 1996).