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FREEDOM AND RESPONSIBILITY IN GENETIC TESTING

By Baruch A. Brody

I. Introduction

Public statements by various international groups emphasize that decisions to undergo genetic screening, either for disease-carrier status or for predisposition-to-disease status, and decisions about the use of the resulting information should be made voluntarily by the party to be screened. For example, the World Medical Association, in its Declaration on the Human Genome Project, says, "One should respect the will of persons screened and their right to decide about participation and about the use of the information obtained."[1] Giving this principle a name, "voluntarism," the Council for International Organizations of Medical Sciences, in its Declaration of Inuyama (the host city for the conference), announced that "voluntarism should be the guiding principle in the provision of genetic services."[2]

In part, the adoption of this principle represents an attempt to distinguish the "new genetics" from the "old eugenics." The old eugenics was a movement in the United States and Great Britain in the latter part of the nineteenth and the first half of the twentieth centuries that emphasized "genetic improvement" of the population. It was criticized, at least in part, because it forced decisions on people—for example, forced sterilization. The new genetics, by contrast, is thought to be acceptable precisely because it merely offers people opportunities to obtain genetic information through screening, information which they can then use as they see fit. In part, the adoption of this voluntarist principle also represents a commitment to general principles of autonomy, which require us to respect the decisions of individuals as to whether or not certain things may be done to them.

In this essay, I want to argue that the voluntarist approach fails to confront a number of crucial issues about both freedom and responsibility that are raised by the new genetics. In particular, I want to consider (1) issues of reproductive responsibility as well as reproductive freedom, and

[1] World Medical Association, "Declaration on the Human Genome Project," in World Medical Association, *Handbook of Declarations* (Ferney-Voltaire, France: World Medical Association, 1995), 17.

[2] Council for International Organizations of Medical Sciences, "Declaration of Inuyama," in Zbigniew Bankowski and Alexander M. Capron, eds., *Proceedings of the 24th CIOMS Round Table Conference* (Geneva, Switzerland: CIOMS, 1991), 1–3.

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(2) issues of unconstrained and responsible access to available genetic testing.

II. Reproductive Responsibility

Suppose that there are genetically determined diseases:

(a) which result in the inevitable death of young children before they reach the age of autonomy;
(b) which produce such pain and suffering that it is reasonable to suppose that the child's life until it dies is not a life worth living;
(c) for which relatively cheap and painless genetic tests are available that identify carriers of the defective recessive genes which produce the diseases in question; and
(d) which are such that children will not be born with these diseases if adults with reproductive potential are screened and if, in all cases where two partners are carriers, the pair (i) do not procreate; (ii) procreate only by use of in vitro fertilization (IVF), preimplantation zygote screening, and disposal of affected zygotes; or (iii) procreate only with an understanding that all fetuses will be tested and that those that will develop the disease will be terminated.

If all of these conditions are satisfied, then it seems to me that everyone has a moral responsibility to undergo genetic screening and to adopt one or more of the strategies outlined in (d)(i)–(iii). Alternatively, if there are special groups that have a serious risk for these diseases and in which most of these diseases are confined, all members of those groups have a moral responsibility to undergo genetic screening and to adopt one or more of the strategies outlined in (d)(i)–(iii). Let me call this *the moral responsibility to be at least minimally responsible in reproduction*.[3] The basic justification of this claim of moral responsibility is the idea that producing a child who has a terminal genetic disease that causes him suffering is harming that child, and one has an obligation not to harm nonconsenting others by taking measures to avoid serious chances of producing such harm. After explaining and defending this view more fully, I will turn

[3] In asserting the existence of this moral obligation, I am by no means suggesting that there are not additional moral obligations in the area of procreation, responsibilities related to not producing offspring that would have very poor lives, although lives that would on the whole be worth living. This is why I have labeled the obligation in the text the obligation to be "minimally responsible." The exact nature of those additional moral obligations, and the question of the legal enforcement of them, is being explored by Janet Malek in a doctoral thesis that is being completed at Rice University. Given that the lives in question are lives that would be worth living, the argument in this essay cannot simply be extended to those sorts of cases. Malek is exploring the more complex arguments that are involved there.

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to the question of what should be the social response to this moral responsibility.

It is worth noting that both conditions (a), concerning the shortness of the life, and (b), concerning the poor quality of the life, are satisfied by the diseases I am discussing. In light of the basic justification of the claim of moral responsibility, the mere satisfaction by some disease of condition (a) is not, I believe, sufficient to justify the claim of minimal moral responsibility in reproduction vis-à-vis that disease. If a life in question is a life worth living, but will be a short life, a person is not necessarily harmed by being created. Of course, in assessing whether the life is worth living, one must take into account the impact of the knowledge of early demise. Thus, in the case of cystic fibrosis, my principle of minimal responsibility in reproduction would probably not have applied in past decades, even if a prenatal screening test had been available and even when the median life expectancy of those affected was ten to twenty years, much shorter than it is today, when medical advances have prolonged life. But it is arguable that the mere satisfaction by some disease of condition (b) is sufficient to justify the claim of minimal moral responsibility, because if the life is a life not worth living, then the harm is even greater if the life lasts for a longer period of time rather than a shorter period of time. Conditions (a) and (b) are both listed in my analysis only because those diseases which clearly satisfy (b) seem to also satisfy (a). But the argument rests on the satisfaction of (b), and the mere satisfaction of (a) is not necessary for the claim of minimal moral responsibility to hold.

This line of argumentation has recently been developed by Allen Buchanan and his colleagues.[4] There is a lot of justification required to complete the argument, including the defense of three important claims: (1) that there are lives that are not worth living, (2) that someone is harmed by being created to live such a life, and (3) that the harm in question is not one which people are entitled to impose upon others. I would note that while I agree with much of what Buchanan has to say about claims (1) and (2), I want to emphasize the need to defend more fully claim (3). In competitive situations, for example, we often are entitled to impose harms on others when pursuing important interests of our own. Some might argue, appealing to the important interest in reproductive freedom, that this applies to conception issues as well. They might argue that our interest in being able to have biologically related children with whomever we want, to do so without using techniques of assisted reproduction, and

[4] Most recently, in Allen Buchanan et al., *From Chance to Choice: Genetics and Justice* (New York: Cambridge University Press, 2000), chap. 6. In our agreement on this approach, we differ from David Heyd's position of "generocentrism," which is the position that "genesis choices should be guided exclusively by reference to the interests, welfare, ideals, rights and duties of those making the choice, the 'generators,' the creators, or the procreators." This alternative view is announced in David Heyd, *Genethics* (Berkeley: University of California Press, 1992), 96, and is defended in Part II of that book.

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to do so without terminating affected pregnancies is so important that it entitles us to impose harms on the children who are created. The important interest in reproductive freedom might serve, then, as the basis for an objection to the postulated moral obligation to be minimally responsible. While such an objection might be raised, I find it implausible for two reasons. First, in a competitive situation, we are harming strangers to whom we bear no special obligation to not harm. This is why one needs to identify another reason (e.g., we violate one of their rights in some cases by harming them) in order to justify claims that we are not entitled to impose particular harms on strangers. In the procreation case, by way of contrast, the child created is one to whom the creators have special obligations. Those special obligations to one's children are usually understood to comprise more than the obligation of not harming them, but it certainly includes this obligation. This obligation serves as the reason why one is not entitled to harm them, even given one's significant interests in procreation. Second, it is important to note that each reproductive interest (i.e., being able to have biologically related children with whomever we want, being able to do so without using techniques of assisted reproduction, and being able to do so without terminating affected pregnancies) can be satisfied, even while the obligation to be minimally responsible in reproduction is met and the child produced is not harmed. It is only the joint satisfaction of all three interests that poses a problem for not harming the child who is created. Arguably, the interest in jointly satisfying all three is less important to parents than the satisfaction of each (or any two).

This last point is related to the wide range of ways, indicated in (d), by which the postscreening portion of the obligation to be minimally responsible in reproduction can be met. This wide range is very important for another reason. It means that there will be techniques available to nearly everyone for meeting the above-postulated postscreening ethical obligation, regardless of their views about a wide variety of contentious moral issues. For those whose views allow for the termination of pregnancies in utero once devastating fetal testing results are in, option (d)(iii) is available. For those whose views only allow for termination of pregnancy before implantation and also allow for use of IVF, option (d)(ii) is available. For those whose views oppose all terminations whether in utero or in vitro, a form of option (d)(i), keeping the partner but not procreating, is available, unless they also oppose contraception (artificial and natural) and/or insist that there is a moral requirement to produce genetically related children. Finally, for those who oppose all of the above, another form of option (d)(i) is available, choosing a different reproductive partner, and has in fact been adopted in some very traditional religious communities. The important point is that nearly every person seems to have available at least one way of meeting the above-postulated postscreening moral responsibility that is compatible with his or her other moral beliefs

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about such matters as reproduction, contraception, and abortion.[5] One cannot object, then, to the obligation to be minimally responsible in reproduction on the ground that it calls upon people to violate other crucial moral beliefs.

Another possible objection that might be raised to the claim that there is such an obligation is that its recognition involves an inappropriate stigmatization of disabled people and a failure to appreciate the value of their lives. This objection is related to a criticism levelled against the old eugenics: that it was wrong not just because it imposed eugenic measures, such as sterilization, on people against their wills, but because it failed to appreciate the value of the lives of those disabled by birth defects. This failure resulted in a stigmatization of those living with disabling conditions. While this is a perfectly appropriate assessment of the old eugenics, it does not apply to our postulated minimal moral obligation because of the stringency of condition (b). Whereas the old eugenics supports the claim that it is irresponsible to produce mentally or physically handicapped children (e.g., children with Down's syndrome or children with spina bifida), our principle will not, because condition (b) is not satisfied. Only the most devastating diseases, such as Tay-Sachs disease or Lesch-Nyhan disease, raise this obligation. These diseases involve so much pain and suffering before death with so little in compensating benefits that those suffering from them are properly described as having lives not worth living.[6] The postulated obligation rests upon an appropriate assessment of the value of these lives, and it is not stigmatization to describe lives not worth living as being such.

Those who object to genetic screening on the ground of stigmatization might respond that there are two other groups of people being stigmatized: those who turn out to be carriers and the subgroups in which they predominate. Thus, in the case of Tay-Sachs disease, even if there is nothing inappropriate in saying that the lives of the children with the disease are not lives worth living, we still harm those who test positively as carriers. They are stigmatized as being "dangerous" to choose as partners with whom to reproduce. Moreover, the general results of the testing

[5] The most stringent of the positions, that is, the one that must fall back on the choice of the different reproductive partner, would meet even the demands of traditional Roman Catholic moral theology, which would not allow for termination of pregnancy either in utero or before implantation and would not accept artificial contraception. For an analysis of these options in the most traditional segments of the Orthodox Jewish community, where the issue of Tay-Sachs screening to help choose a marriage partner has been very important, see J. David Bleich, *Judaism and Healing* (New York: Ktav Publishing, 1981), 103–9.

[6] Tay-Sachs disease involves a destruction of neurons. After four to six months of normal development, the child begins to deteriorate, developing poor muscle tone and a loss of interest in his surroundings. Seizures begin at a later stage. In advanced stages of the illness, there is no response to external stimuli. Lesch-Nyhan disease, which is due to abnormalities in uric-acid metabolism, is even more striking, as it involves compulsive self-destructive behavior, such as biting and chewing fingers and lips, even though this is clearly painful to the patient.

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stigmatize the Ashkenazic Jewish community as being a community that is inferior because of the disproportionate prevalence within it of a terrible disease.

The last point, about stigmatization of the community, is clearly in error. The disproportionate prevalence exists and is known independently of any genetic screening for carrier status. It is not the screening which poses the problem for the community. If anything, it offers the community an opportunity to demonstrate its moral responsibility by developing and carrying out a model screening program. The problem concerning those individuals who are stigmatized because they test positively as carriers is a more serious issue. Against this problem, I would raise a number of considerations: First, for those for whom there is a known family history of the disease and who test negatively as a carrier, screening is actually a benefit. Second, even those who test positively pose no threat as reproductive partners to the overwhelming majority of people, since most others test negatively, so that mitigates the stigmatization. Third, as shown above, even two carriers have options for responsible reproduction. None of this is to deny the existence of residual stigmatization, but this has to be balanced against the prevention of serious harm, and it does not seem that fear of residual stigmatization is sufficient to override the moral obligation to be tested.

What I now want to consider is the question of permissible social policy in response to this moral obligation. Obviously, the mere recognition of a *moral* responsibility to undergo certain genetic screening tests and to act in the ways indicated above does not in any way impinge upon the voluntariness of the decision to be screened. Moreover, autonomy does not mean making decisions while being unaware of one's moral obligations; in fact, it may mean just the opposite. Despite the fact that moral obligations do not undermine voluntariness or autonomy, the issues of voluntariness and autonomy do come to the forefront when one considers social policies that might require fulfillment of the moral obligation I have been discussing.

The first point that needs to be made is that any such policy would not violate John Stuart Mill's "harm principle."[7] This principle affirms that a necessary condition for the legitimacy of the use of the coercive powers of the state to prevent certain behavior is that the behavior would produce a harm to which no one consented. The very justification of the claim that there is a moral obligation to be minimally responsible in reproduction is that the failure to do what it postulates harms unconsenting victims (in fact, given that the victims are children, they cannot consent). We are not dealing here with paternalistic or moralistic uses of the coercive powers of the state, which are uses of the powers of the state, respectively, to protect the person performing the action or to prevent immoral actions

[7] John Stuart Mill, *On Liberty* (Indianapolis, IN: Hackett Publishing, 1978), 9.

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that do not have victims. But that of course does not settle the issue, even for followers of Mill. The satisfaction of the harm principle is at most only a necessary, not a sufficient, condition for the legitimacy of coercive state action. It remains to be considered whether a sufficient justification can be found to use coercive state action to enforce the moral obligation to be minimally responsible in reproduction.

In considering this challenge, it is important to differentiate between a legal requirement to be screened for carrier status (legally compulsory screening) and a legal requirement to adopt one of the measures in (d)(i)–(iii) in response to a positive test result (legally compulsory postscreening responsible reproduction). The moral obligation may be to do both, but there may be reasons to legally require the former but not the latter. We shall consider such reasons below. It might be supposed that there is no point in legally requiring testing for carrier status if there is no further legal requirement to do something in response to a positive test result. I would reject that supposition. It seems quite reasonable to suppose that there are people who would not get tested unless they were required by law to do so, but who would act responsibly if they were tested and shown to be carriers, even without any legal requirement to do so. Before one knows that one is a carrier, the possibility of harming the children that one will create seems so remote that one might not be tested, even if one acknowledged a moral obligation to be tested. After one knows that one is a carrier, that possibility becomes very personal and very real, and no further legal coercion may be required to produce minimally responsible reproductive behavior. We need, then, to examine separately whether there are any reasons why legally compulsory screening for carrier status should not be introduced and whether there are any reasons why legally compulsory postscreening responsible reproductive behavior should not be introduced. One might not introduce legal compulsion in either case because of practical reasons or because the compulsion introduces additional moral problems. Both of these factors need to be considered separately for the screening and the postscreening legal compulsions.

Compulsory screening for carrier status faces at least three practical issues: At what point should the requirement be imposed to make it reasonably enforceable? Is it financially feasible? What counseling should be provided about the meaning of the results? This is not the place to examine the data (such as there is) relevant to these three questions. So let me just say how I would start to address them. It would be best to impose the requirement of being screened as a condition for receiving a benefit wanted by most teens at a time when they are just beginning their sexual activities. Thus, imposing it as a requirement for obtaining a driver's license rather than a marriage license would be preferable. While receiving a driver's license is not related to reproduction, I see no reason to insist that there be a linkage. The cost concerns could be mitigated by a single inexpensive test, which would test for carrier status for a full

battery of genetic diseases. New technologies (such as interactive computer programs) could be helpful in postscreening counseling. All of these points need to be developed further. Special attention would need to be paid to the issue of the social and psychological implications for teenagers of being screened. But I think that these practical concerns can be adequately addressed.

These practical concerns are not, however, the primary issue. The primary issue is whether making screening for carrier status legally compulsory, if such screening is morally obligatory, is ruled out by other moral considerations related to compulsion. Such considerations include: that it is wrong because it is compulsory; that it is wrong because it is a compulsory invasion of one's body; or that it is wrong because the compulsion violates one's reproductive freedom.

The first of these considerations is not promising. No doubt, legally compulsory screening for carrier status is—by definition—compulsory, but the whole issue before us is whether it is justifiable compulsion. The serious harm to nonconsenting others it is designed to prevent gives us a prima facie reason for thinking that the compulsion is justifiable. We are looking for some additional consideration as to why it is not in the end morally justifiable. Merely to be reminded that it is compulsory gives us no such additional consideration.

The second consideration attempts to do just that. It reminds us that the compulsion in question is of a specific type: a bodily invasion. Bodily integrity (in the sense of our bodies not being compulsorily invaded), it might be suggested, is a crucial right, and its existence argues against compulsory genetic screening. It is just this sort of consideration which has been offered, among others, against compulsory sterilization or compulsory abortion in the eugenic setting, or against compulsory health care more generally. While more promising, this second suggestion does not in the end prove telling. This consideration carries far more weight in the cases of compulsory sterilization, abortion, and health care than in the case of compulsory genetic screening for carrier status, as genetic screening is certainly less invasive and might not be invasive at all if it were performed on biological material already separated from one's body—for instance, hair. This does not mean that individuals have no claims against even minimally invasive bodily invasions or against the nonconsensual use of separated bodily material. It just means that these claims have less moral weight than the claims against the bodily invasions involved in nonconsensual sterilization, abortion, and many forms of health care, and that the former could well be outweighed by the social need to protect against the harm of producing lives not worth living. We already have examples of such legitimate, compulsory, minimally invasive bodily invasions and/or compulsory use of separated bodily material in a variety of public health measures designed to detect infectious diseases and to prevent their spread (e.g., compulsory testing and treatment for tuberculosis).

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It is important to note that such programs exist and can be justified. In all cases, however, every effort should be made to ensure that private information does not become public; it is important to protect confidential information.

The third consideration, the issue of reproductive freedom, is not relevant at this point in the dialectic.[8] There is no point in raising this consideration as an appeal to a legal right of reproductive freedom, since the very question we are considering is whether any such legal right should include the right not to be tested for carrier status. Nor will an appeal to a moral right of reproductive freedom do any better. Suppose, as we do at this point in the discussion, that it has been established by the arguments above that the moral obligation to be at least minimally responsible in reproduction does exist, along with the moral obligation to be screened for carrier status. If we suppose this, we must be defining the moral right of reproductive freedom in a way that does not include the moral right to avoid being screened. One cannot, then, invoke the moral right of reproductive freedom as the basis for a moral argument limiting state compulsion to help prevent the harms produced by people who fail to be screened.

All of this looks very different, of course, when we turn from the issue of legally compulsory genetic screening for carrier status to legally compulsory postscreening responsible reproduction. Both the practical problems with, and the moral constraints on, the use of legal compulsion become far greater for the latter.

The first point to be noted about legally compulsory postscreening responsible reproduction is that such use of legal compulsion would have to be very different from the legal compulsion to be screened for carrier status. In screening for carrier status, as I envisage it, the state would compel the performance of a one-time, desirable behavior by withholding an expected benefit until the behavior was performed. This policy would be practical while avoiding serious moral problems. But if the state were to use compulsion to prevent the performance of irresponsible postscreening reproduction over the reproductive lifetime of an individual, it is hard to see how this could be done except through the threat of punishment. Direct attempts to prevent such behavior—by preventing sexual relations with reproductive potential between partners who are both carriers, by

[8] It is, nevertheless, often invoked as though it settles the question. Thus, the American Society of Human Genetics, in a statement entitled "Eugenics and the Misuse of Genetic Information to Restrict Reproductive Freedom," states that it "deplores laws, government regulations, or any other coercive effort intended to restrict reproductive freedom or constrain freedom of choice on the basis of known or presumed characteristics of potential parents or the anticipated genetic characteristics, health or capacities of potential offspring." American Society of Human Genetics, "Eugenics and the Misuse of Genetic Information to Restrict Reproductive Freedom," reprinted in Baruch Brody et al., *Medical Ethics: Codes, Opinions, and Statements* (Washington, DC: Bureau of National Affairs, 2000), 245. Oddly enough, the same group, in that same statement, approves of laws forbidding first-cousin marriages.

compelling abortions of affected fetuses, or by preventing implantation of affected zygotes—would involve serious invasions of both privacy and bodily integrity that would be both impractical and morally unacceptable.

The second point to be noted is that the state has alternatives. It can encourage postscreening responsible choices by educating affected couples about adoption and streamlining the adoption process, by paying for voluntary testing of pregnant women and voluntary terminations of affected fetuses, or by paying for assisted reproduction with preimplantation genetic testing of zygotes. All of these measures, combined with compulsory screening for carrier status and follow-up counseling for those who are carriers, may provide an alternative way to avoid the harm of bearing children who have severe genetic diseases. If more is needed and if compulsion is to be avoided, there is always the option of financial incentives to reward minimally responsible reproductive behavior on the part of those who are carriers. Defending this latter option requires, of course, being able both to differentiate such incentives from compulsion (by arguing that offers, in general, or these offers, in particular, are not compulsive or are not as compulsive as the threats of punishment for irresponsible behavior) and to defend such incentives against other moral charges (e.g., exploitation or commodification). While this essay is not the place to attempt either of these tasks, I do think that this other option for state action needs at least to be mentioned.[9] These alternatives to the use of threats of punishment may well be preferable either because they are more likely to work or because they are morally preferable since the harm can be avoided without the use of postscreening compulsion.

In summary, I have argued in this section that there is a moral obligation to be at least minimally responsible in reproduction. I have also argued that screening for the carrier-status component of that obligation might appropriately be compelled by the state through a well-designed program, but that the postscreening component of that obligation is better promoted through incentives and facilitation rather than through the use of compulsion.

III. Access to Available Genetic Testing for Disease Predisposition

In Section II, I considered the question of whether people who do not want to undergo certain genetic testing have a moral obligation to be

[9] Good background discussions of the issues related to coercion and exploitation are found in Alan Wertheimer, *Coercion* (Princeton, NJ: Princeton University Press, 1987); and Alan Wertheimer, *Exploitation* (Princeton, NJ: Princeton University Press, 1996). Margaret Radin's *Contested Commodities* (Cambridge, MA: Harvard University Press, 1996) is an important general introduction to the issues of commodification. I know of no attempts to analyze the issues of coercion, exploitation, and commodification in work related to financial incentives for encouraging minimally responsible reproductive behavior.

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tested which they might appropriately be coerced into fulfilling. But might there be a different type of case in which people want to undergo genetic testing but no one will provide them with it even if they are willing to pay for it? And if there is such a case, do people, in their exercise of responsible freedom, have a right to be tested? Do the principles of voluntarism and respect for the choices of individuals with which this essay began require that they be tested by those who have the capacity to do the testing?

This is not just a theoretically possibility. An actual example is available in the area of genetic testing for a predisposition to develop Alzheimer's disease. While the exact scientific details need further elucidation,[10] it is clear that there is an association between the APOE4 genotype (especially a homozygous genotype) and a greater likelihood of developing Alzheimer's disease. But this is only one of the risk factors for that disease. Many (perhaps a majority) of those who develop the disease do not have that genotype, and many (perhaps a majority) of those who have that genotype do not develop the disease. Using the presence of this genotype as a predictive test for developing the disease, then, is neither a very sensitive nor a very specific test. In addition, since there are at the moment no known effective preventive measures or early treatments which delay the progression of Alzheimer's disease, knowing whether you have this genotype gives you no medically useful information. Moreover, knowing that you do have this genotype, especially if the information is misunderstood (but even if it is properly understood), may have a negative psychological impact, primarily by provoking unnecessary anxiety. Keeping all of this in mind, a variety of professional groups have argued that genetic testing for Alzheimer's susceptibility should not be made available, and they have criticized companies that plan to do so.[11] The issue is complicated by the fact that the same testing may be valuable for the management of cardiovascular disease, since that same genotype is implicated. How to use the same test for cardiovascular purposes without indirectly using it as a screen for susceptibility to Alzheimer's disease is unclear.

A preliminary analysis might suggest that none of this is problematic. After all, clinicians, as well as patients, have a right to have their responsible, autonomous choices respected. So when there is a conflict between a patient's choice and a physician's choice, this is a conflict between two parties with equal rights to respect for their responsible, autonomous

[10] A useful summary of the literature is found in Stephen Post et al., "The Clinical Introduction of Genetic Testing for Alzheimer Disease," *Journal of the American Medical Association* 277, no. 10 (1997): 832–36.

[11] See, for example, American College of Medical Genetics/American Society of Human Genetics, "Statement on Use of Apolipoprotein E Testing for Alzheimer Disease," *Journal of the American Medical Association* 274, no. 20 (1995): 1627–29; and National Institute on Aging/Alzheimer's Association Working Group, "Apolipoprotein E Genotyping in Alzheimer's Disease," *Lancet* 347, no. 9008 (1996): 1091–95.

choices. Normally, when the physician wants to do something to the patient (e.g., run a genetic test) which the patient does not want done, the patient's right to have his choice respected takes precedence, primarily because of his supporting right of bodily integrity.[12] But when it is the patient who wants something done to him (e.g., to be genetically tested for disease susceptibility) that the physician does not want to do, it is the physician's right to have his choice respected that takes precedence, primarily because of the physician's supporting right to not have a positive servitude imposed upon him. This is especially true when the refusal to provide the service is based upon the physician's judgment that the service requested is inappropriate, because the physician's right to preserve his personal and/or professional integrity then plays a supporting role as well. An important additional question, which lies beyond the scope of this essay, is whether anyone else (e.g., the system in which the physician is working) has an obligation to provide the requested service.

I do not want to challenge this preliminary analysis as incorrect. Rather, I want to suggest that it is incomplete because it fails to call upon the physician to reassess the reasons for his refusal to respect the patient's choice to be tested in light of a better understanding of the reasons for respecting the responsible, autonomous choices of a patient.

Why should a physician respect the responsible, autonomous choices of patients? One reason is given in the above rights-based analysis. It is the right of bodily integrity that supports respecting a patient's refusals of interventions. This obviously applies only to refusals of interventions, as the above analysis suggests. But this is not the only reason for respecting a patient's decisions. There is also the general point, suggested by Mill, that the individual in question is the best judge of what is beneficial to him.[13] Given that this is so, the physician who wishes to help the patient should respect the patient's choice because doing so is likely to be beneficial to the patient. This type of reasoning applies to requests for interventions as well, and has a major impact on the above rights-based analysis. In those cases in which the physician's refusal is based upon his judgment that the requested service is not beneficial to the patient, the physician, rather than insisting on his right not to have a positive servitude imposed

[12] There has been considerable legal discussion about whether informed consent must be obtained by a physician only for providing treatment (where there would otherwise be a violation of bodily integrity) or also for not providing treatment (where the failure to provide treatment certainly does not involve any violation of bodily integrity). The adoption of the second position is compatible with my claim in the text. This legal debate is about whether physicians must provide certain information to patients to enable the patients to make an informed choice, even when what the physicians want to do involves no violation of bodily integrity. This is a very different question from the question of whose choice takes precedence when choices are in conflict, which is the question I am discussing in the text. There may be value in the patient's receiving the information and making a choice, even if the patient cannot force the physician to follow that choice. If nothing else, it lets the patient know that he may need to find another doctor who will be willing to follow his choice.

[13] Mill, *On Liberty*, 81–82.

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upon him, might instead reassess his refusal to provide the requested test and conclude that it should be provided because it is probably beneficial to the patient. At least, he might do so after a full exploration of the issues with the patient and after assuring himself that the patient's request is not based upon a misunderstanding or pathology. Naturally, this alternative approach would not apply in other cases in which the physician's basis for rejecting the patient's request is based on different considerations, ranging all the way from moral opposition to what is being requested of him to the patient's inability to pay for the services requested.[14] Let me illustrate this approach by returning to the case of the patient requesting APOE testing to determine genetic susceptibility to Alzheimer's disease.

The first thing we need to do is determine the reasons typically given for rejecting a request for genetic testing of this type. An analysis of the reasons offered by professional groups against APOE testing for Alzheimer's makes clear what our patient would likely hear from a recalcitrant physician. The professional groups argue purely on the ground that being tested is not beneficial to the requester. Three main arguments have been offered and they are all of this nature:

(A) *The genetic tests are neither specific enough nor sensitive enough.* People who test positive for the homozygous APOE4 genotype may conclude that they are going to develop Alzheimer's disease. This is in error, though, because many (perhaps the majority) of those who test positive will not develop the disease. Similarly, those who test negative for that genotype may conclude that they are not going to develop Alzheimer's disease, but this too is in error because many (perhaps the majority) of those who develop the disease do not have this genotype. The risk that these erroneous conclusions will be drawn persists even after appropriate pretest and posttest counseling. It is not beneficial to anyone to provide a person with information that may be misunderstood.[15] Therefore, we should not provide people with information about their APOE genotype.

[14] Much more needs to be said, but not in this essay, about whether physicians should exercise their rights in these other cases. In the case of conflicts of conscience, this relates to questions of complicity that are being explored by J. Clint Parker in a thesis being completed at Rice University. In the case of inability to pay, this relates to questions of whether the patient has a right to that care, and if so, whether the particular physician has any special obligation to see that the patient's right is fulfilled.

[15] This general claim seems to be presupposed by the argument as usually presented. It is, of course, an absurd claim, since there always is a possibility that information can be misunderstood. Some modification is required. One thought is that one confine the claim to when there is a very significant possibility of the information's being misunderstood and when that misunderstanding can lead to serious losses for the party misunderstanding the information. But then the proponents of this argument in connection with APOE testing will need to show that these additional assumptions are true in that case.

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(B) *The information provided by the tests is medically useless*. People who test positive for the homozygous APOE4 genotype cannot do anything medically useful with that information. They know that they are at higher risk for developing Alzheimer's disease, but there are no known preventive measures that they can take to lessen that risk, nor are there any useful treatments available to slow disease progression before the disease becomes symptomatic. This differentiates APOE genetic testing from genetic testing for cardiovascular disease or for breast cancer. It is not beneficial to anyone to provide a person with useless information. Therefore, we should not provide people with information about their APOE genotype.

(C) *The tests provide psychologically burdensome information*. People who test positive for the homozygous APOE4 genotype are at great risk for excessive anxiety (every lapse in memory is interpreted as the beginning of disease onset) and for depression (their future is hopeless). These psychological burdens of living with this type of information make it difficult to live a normal life in the healthy years before the disease develops, if it ever does develop. It is at least in part for this reason that so many people with a family history of Huntington's disease have refused to be tested, preferring to live with uncertainty.[16] It is not beneficial to anyone to provide a person with information that produces this type of psychological burden. Therefore, we should not provide people with the information that they are APOE4 homozygous.

To be more accurate in our analysis of these arguments, we should recognize them as not being independent of each other. If there were real benefits from having the information accurately understood, these benefits might outweigh the psychological burdens. Each of these arguments is supplying only a prima facie reason for seeing APOE genotype testing as not being beneficial to the party wanting to be tested. Together, they represent an all-things-considered argument for this claim.

The next thing to do, given these reasons by professional groups for not respecting requests to be tested, is for a physician to reassess these reasons in light of a particular patient's perception that it is beneficial to him to be tested. Suppose that the following patient presents his physician with a request to be tested: the patient makes it clear that he understands the lack of test sensitivity and specificity and recognizes that the most he will be learning is whether he has an increased susceptibility to developing Alzheimer's disease. He also recognizes the increased risk of anx-

[16] Marlene Huggins et al., "Ethical and Legal Dilemmas Arising during Predictive Testing for Adult-Onset Disease: The Experience of Huntington Disease," *American Journal of Human Genetics* 47, no. 1 (1990): 4–12.

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iety and depression if he tests positively, but he thinks that these risks are manageable. Moreover, he sees two offsetting benefits in his life. One is that he believes that the information will be useful to him in planning his life. If he is at increased risk, he may make an effort to accomplish certain goals (e.g., extensive foreign travel) earlier in his life, since he may not be able to depend upon doing so in his later years. The other is that a negative test result may offer some reassurance to him, as he recognizes that he is already concerned about the prospect of developing this disease. Others may not see these benefits as sufficient to outweigh the risks, but he thinks that they are sufficient *for him*. If we really believe that informed and responsible individuals are the best judges of what is beneficial to them, and we see that as a reason for respecting their responsible, autonomous choices, then why shouldn't the physician conclude that he should test this patient because the best judge has concluded that it is beneficial to him?

As indicated above, the physician may think that he should probe more fully the patient's understanding of the issues in question. As part of that process, he may want to explain the arguments against testing and see what the patient says in response. This might result in the patient's really changing his or her mind, and there is nothing wrong with that. The physician may also want to assure himself that there is no hidden pressure from someone else (e.g., the patient's spouse or children) that led to the request for testing. But if the physician is satisfied after his counseling and probing, why shouldn't he provide the test? No doubt, he is within his rights to refuse to have this positive servitude imposed upon him, but he should not justify his refusal on the ground that his refusal is beneficial to the patient. If he has no other justification, why should he insist upon exercising his right to refuse? We should not always insist upon exercising our rights just because we have them.

Should we conclude, then, that physicians should make APOE genotype testing available to patients requesting it, provided that they have done the above-described counseling? I am not sure that the analysis offered so far is sufficient to justify that conclusion, for more needs to be said about the meaning and justification of Mill's claim, upon which this analysis is based, that the patient is the best judge of what is beneficial to him.

When we ask whether a certain action is beneficial to someone, we need to consider both what the interests of the party in question are and how the action will affect those interests.[17] Only an answer to both of

[17] The same point needs to be made in connection with the "corrected preferences" theory of interests that are defined in terms of preferences. It is one thing to discount preferences for the satisfaction of certain goals that are based upon false beliefs that the satisfaction of those goals will lead to the satisfaction of other, more fundamental preferred goals. It is a very different thing to discount preferences for the satisfaction of fundamental goals on other grounds.

those questions will settle the issue. Is Mill's claim true about the answer to both of these questions?

Mill's answer has greater plausibility when addressed to the question of what are the patient's interests, because we see different individuals as having different interests. We might suppose that different individuals have different interests because we accept some version of the preference-satisfaction theory of interests and we recognize that people have different preferences. But even if we accept a more objective account of an individual's interests, we are still likely to see that different individuals have different interests, because many of the usual items on the objective list (e.g., feeling a sense of meaning and accomplishment) have a subjective component (since a result which makes one person have those feelings may not produce those feelings in others). Unless we adopt a narrow objective theory of interests, which allows no room for this type of variability, we will conclude that different individuals have different interests. Given that a narrow objective theory of interests is unpersuasive, it then seems plausible to go on to say that individuals are the best judge of what their interests are. It is important to note that this further conclusion, while plausible, is not necessarily true. It depends at least in part upon how much people are aware of what they really care about. If there are real differences between what people think they care about and what they really care about, then they may not be such good judges of what their interests are. But I am inclined to accept this further conclusion anyway because of its comparative component: while a given individual may not be fully aware of what he really cares about, he is likely to be more aware than anyone else.

This type of reasoning is not so easily transferable to the question of how the action in question will affect the individual's interests. Let us grant that different people will be affected differently by a given action. Let us further grant that this means that the interests of different people will be affected differently by a given action, even if the same interests are at stake. Is it plausible to suppose that the individuals are the best judge of how they will be affected by a given action? The individuals in question bring their knowledge of how they have been affected in the past by such actions. Outside experts bring their knowledge of how people in general have been affected by such actions. Which knowledge is more valuable? The more the individual has had similar experiences and the more people vary in the effect of the action upon them, the more valuable is the individual's own knowledge. The more people's responses to such actions are similar and the less the individual has had similar experiences, the more valuable is the knowledge of the experts. In response to this second question, then, Mill's answer may be wrong.

Let us return to our Millian analysis of APOE genotype testing. We should probably grant that the individual is the best judge of what are his interests that might be affected by the testing, since it will be a rare

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physician who knows his patient well enough to claim that he knows better than the patient what the patient cares about (e.g., the importance in his life of extensive foreign travel). But what about the question of how the individual and his interests will be affected? Who is the better judge of that? Who knows better whether the patient will draw excessive conclusions or just recognize that he has acquired information about susceptibility to developing a disease? Who knows better whether the patient can manage the possible anxiety or depression? Given the above criteria, neither knows very well. The knowledge of the experts is limited because of the admitted variability in how people are affected by obtaining knowledge of genetic susceptibility. The knowledge of the individual is limited by the limited past experience he has with acquiring such knowledge. If we conclude that the physician should respect the patient's request for APOE genotype testing on the ground that the patient is the best judge of what is beneficial to him, it should not be on the ground that the patient is necessarily a very good judge, but merely on the ground that he is likely to be a better judge than his physician.

The conclusion of this section is, therefore, more tentative than the conclusion of Section II. Individual physicians are certainly acting within their rights if they refuse to provide problematic tests of disease susceptibility, such as APOE genotype testing, to patients who request them. But if, as seems to be the case, their reasons for doing so are their concerns about what is beneficial to the patient, then they may well be advised to provide the testing, after appropriate counseling, to the patient who requests it, since the patient requesting testing is probably a better judge.

IV. Conclusion

The principle of voluntarism in genetic testing is an initially attractive principle. More careful reflection has shown, however, that it needs to be modified in at least several ways. It needs to take into account types of genetic testing that individuals are morally required to undergo, and that may even be justifiably mandated. It needs to take into account the extent to which the individual's choice to be tested can generate, in at least some cases, requirements on reluctant physicians to provide tests. A consideration of other examples may indicate additional ways in which the principle of voluntarism should be modified. In a moral universe in which many values need to be balanced, it is a mistake to try to ground the ethics of genetic testing in one simple principle.

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