2001

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Hard Cases for Autonomy, Respect, and Professionalism in Medical Genetics*

Roger B. Dworkin

Case 1

John, a 54-year-old man, who is at risk for Huntington's Disease (HD), is on the waiting list to receive a heart transplant. John's age-adjusted risk of contracting HD is 10%. The heart transplant center asks the genetic testing center to perform presymptomatic testing to discover whether John will develop HD. If the test is positive, the transplant center will remove John from the waiting list. If the testing center refuses to test John, the transplant center may decide to remove him from the list anyway.

What ought the testing center to do? What is to be said about the transplant center's behavior?

Case 2

We are learning that many conditions not previously thought to be genetic, do have a genetic basis, and that some persons have genetic predispositions to developing still other conditions. As presymptomatic, and especially prenatal diagnosis become more common, difficult questions about how to respond to these conditions will arise.

Suppose it becomes possible to prenatally diagnose alcoholism, homosexuality, and a predisposition to schizophrenia and breast cancer.

1. Mr. and Mrs. A, distressed by the toll that alcoholism has taken on the families of Mrs. A's father and brother, seek prenatal diagnosis to discover whether their fetus will have the mutation that causes alcoholism.

* I am indebted to Kimberly Quaid, Ph.D. for calling case 1 to my attention and to Susan H. Williams for her insightful comments on an earlier draft of this article. A version of this article was presented at a conference, "Hard Cases in Genethics," which was held in Memphis, Tennessee, March 31-April 4, 1999. I am grateful to the organizers of the conference and to its participants for their helpful discussion of the cases and ideas considered in this article.
2. Mr. B. and Mr. C, who are in a committed, monogamous relationship, seek the services of a fertility center to have a child through surrogate motherhood, using semen provided by B or C.

a) The fertility center wants to insist as a condition of its participation that diagnosis be done at the earliest possible moment to assure that any baby born to B, C, and the surrogate not be homosexual.

b) B and C want to insist that diagnosis be done at the earliest possible moment to assure that any baby born to them and the surrogate will be homosexual.

3. Persons at risk for transmitting predispositions to schizophrenia or breast cancer seek prenatal diagnosis of the predispositions in their fetuses.

What are ethically appropriate responses to each of these situations?

Commentary

Both of these cases are designed to test the limits of certain articles of faith in the genetic counseling community, to expose inconsistencies among those articles of faith, and to explore an alternative approach to resolving issues of genetic ethics. I have discussed elsewhere the genetic counseling profession's asserted commitment to patient autonomy and nondirective counseling and its deviation from that commitment when genetic testing of children is involved. I have suggested that respect for all relevant individuals and close attention to professionalism will provide better solutions to ethical issues in genetics. These cases are an effort to test that suggestion in very difficult contexts and to see what solutions that approach offers to concrete dilemmas.

Case I

At first blush this case seems to be about genetic discrimination. It raises the discrimination question in starker, if less common, terms than the usual case about whether to deny insurance or charge extra premiums to persons at risk for developing a genetic disease or to those who have been diagnosed as pre-symptomatic for a genetic disease.

Organs are a very scarce resource. In the United States 10 patients die each day while waiting to receive an organ transplant. As of December 18, 1998, 63,833

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2 http://sdsmarketplace.com/ornfact.htm, crediting statistics to UNOS and TRANSWEB.
persons were on waiting lists to receive donor organs. Thus, the desire to transplant valuable organs to those persons who are most likely to receive long-term benefit from the transplant is easy to understand. How wasteful it would seem to transplant a heart to John only to have John become symptomatic for HD within months of receiving the transplant. HD would significantly shorten John’s life. Before his death it would reduce the quality of his life to the point where he could not recognize the benefit he was receiving from the transplant or engage in the behavior required to protect his transplanted heart. Moreover, transplanting a heart to John will mean denying that heart to another patient who also needs the heart to survive. It is easy to understand the transplant center’s desire to use its scarce resources for patients who will receive the maximum benefit from them.

On the other hand, many patients with heart disease die from conditions unrelated to their heart disease. The transplant center is asking John to pay the price of uneven medical progress, which can now predict whether John will develop HD but cannot predict other maladies or events that may kill other patients. Genetic diseases, like HD, are paradigmatic examples of immutable conditions over which the affected person has no control. If equal treatment means anything, it means that people should not be treated differently because of such conditions. Moreover, requiring John to be tested not only would be to treat him worse than persons who are not (or are not known to be) at risk for HD, but also would be to treat him worse than other persons at risk for HD. Persons at risk are not ordinarily forced to be tested, and the majority of such persons choose not to be. How can one justify forcing John to be tested based on his heart condition, a condition that he may have developed despite his best efforts to pursue a healthy lifestyle?

The case is further complicated by two other facts: First, John’s age adjusted risk of developing HD is only 10%. One wonders whether at some point a risk becomes too low for it to be considered, and, if so, whether 10% is that low. Second, the transplant center may remove John from the list of potential recipients if the genetic testing center refuses to test him as well as if his test is positive. Thus, the only way to assure that John remains on the list is to perform the test and obtain a negative result. The transplant center has placed the testing center, as well as John, in a real bind.

If John refuses to be tested, the testing center obviously cannot proceed to test him. However, John may ask to be tested. He may decide that 10% is a low risk

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3 http://www.giftoflife-sc.org/statistics.asp
4 Ibid.
and that being tested offers him the best chance to receive a transplant. Would it be appropriate for the testing center to refuse to test under these circumstances?

What at first appeared to be a question of discrimination is now revealed to be a question about the limits of professional autonomy: To what extent may genetic professionals refuse to provide services because they disapprove of the reason the services are being sought?

The testing center may want to refuse to test John for at least two reasons: Center personnel may be unwilling to allow the center to be manipulated by the transplant center and to become the passive tool of someone else’s social engineering. Alternatively, the center may believe that testing should only be provided to persons who have freely sought testing or who have sought testing for reasons the center’s personnel approve. These reasons would not justify refusing to test John.

Genetic professionals should be guided by professionalism and respect for individuals affected by the professionals’ work. Nothing in the training or expertise of a genetic professional qualifies him or her to pass judgment on the reasons that a person seeks genetic testing. If the test were medically contraindicated, that would be a different matter. A person who is not at risk for HD should not be tested to allay irrational fears; it is appropriate for a testing facility to conserve its resources. However, a person who is at risk should be able to obtain testing without having to satisfy the nonmedical preferences of the facility and its personnel. John is entitled to the center’s expertise. Its professionals should counsel him about the psychological risks of both positive and negative test results. It is even appropriate to advise him whether to be tested in view of all the matters within the expertise of the professionals. But professionals know no more than anybody else about the intensely personal calculus that will lead one person to prefer a transplant at the risk of acquiring unwanted genetic knowledge and another to prefer genetic innocence and a life shortened by the lack of a transplant. To impose nonexpert judgment on John is simply the unjustified use of power. To do it claiming that it is for John’s own good is arrogance and hubris.

Imposing such a nonexpert judgment on John would also run afoul of the alleged commitment to nondirective counseling. This is not surprising. The commitment to nondirectiveness is often more honored in the breach than in the observance. Genetic professionals are reluctant to advise other people whether to have children,

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6 For the development of arguments supporting directive, rather than nondirective counseling see, ibid., passim; Dworkin, 7 Ann. Rev. of Law and Ethics, op.cit. (fn. 1).

7 This insight underlies the entire law of informed consent. “A medical doctor, being the expert, appreciates the risks inherent in the procedure he is prescribing, the risks of a decision not to undergo the treatment, and the probability of a successful outcome of the treatment. But once this information has been disclosed, that aspect of the doctor’s expert function has been performed. The weighing of these risks against the individual subjective fears and hopes of the patient is not an expert skill. Such evaluation and decision is a nonmedical judgment reserved to the patient alone.” Cobbs v. Grant, 104 Cal. Rptr. 505, 514 (1972).
but they are often willing to impose their other values on patients—refusing to perform presymptomatic diagnosis on children, for example. This apparent anomaly merely reveals the humanness of genetic professionals and the bankruptcy of the nondirectiveness principle. What John and every other patient needs and should be entitled to is the professional expertise of the professional, nothing less, and, as this case suggests, certainly nothing more.

This analysis, however, does not address the testing center’s own interest in not becoming the pawn of the transplant center. If the testing center disapproves of the transplant center’s policy, may it ethically refuse to test John? I think not. John is innocent of any wrongdoing. It seems hard to justify denying him an important benefit to make a point to the transplant center. Moreover, the wisdom and propriety of the transplant center’s policy are beyond the professional expertise of the genetic testers. As citizens, testing center professionals may vote, lobby their legislators, take out ads that criticize the transplant center, etc., but as professionals, they must do what professionals do. Genetic testing professionals exist to do genetic testing in cases where it is consented to and not medically contraindicated. To put it another way, respect for John requires doing what John wants (serving his interest in autonomy) unless matters within the professional expertise of the testers suggest important reasons related to John’s welfare or to the welfare of other identified or reasonably identifiable affected persons to refuse. Refusing to test John because the center feels used by the transplant group is just another way of refusing to test him because his reasons to be tested aren’t good enough. As suggested above, that is not acceptable.

The transplant center’s policy is more difficult to evaluate. The policy is apparently legal as doctors and organ procurement organizations may adopt any criteria they think are appropriate in deciding which patients should be on the waiting lists for organ transplants. As discussed earlier, both the reasons for and against the policy are easy enough to appreciate.

On balance, I think the transplant center’s policy cannot be condemned. The scarcity of organs and the physical and psychological requirements for dealing with transplanted organs are matters within the professional expertise of transplant professionals. Moreover, conditioning a transplant to John on his receiving a negative HD test does not result in an organ being wasted. The organ will be transplanted to somebody else. Any ethical analysis that overlooked the needs of that potential recipient would be incomplete. Each affected individual is entitled to equal respect, and John’s status of being at risk for HD is no reason to prefer John over another potential recipient. Therefore, while the transplant center’s

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policy is not ethically required, neither does it seem to be ethically inappropriate.

Case 2

All prenatal diagnosis cases raise issues about abortion and the genetic testing of children. This is because in the absence of prenatal treatment for a condition, only two outcomes are possible after testing: the pregnancy will be terminated; or a child whose status vis a vis some genetic disease or predisposition is known, will be born. In addition, these cases raise questions of what constitutes a disease, of designing children, and of the limits of parental autonomy.

Schizophrenia and breast cancer are diseases. Alcoholism is widely considered to be a disease, although it is a disease whose symptoms can be completely controlled by the adoption of behavior modification by the affected person. Homosexuality is not a disease, but it is a widely stigmatized minority status. Predispositions are not diseases, although they may be disabilities for purposes of American law. In a sense alcoholism and homosexuality are predispositions in children until they reach the age at which they begin drinking or experience sexual responses.

It is easy to understand why potential parents would prefer to have a child who will not be an alcoholic. Who would prefer a lifetime of struggle and misery for their child and the child’s potential family to a life free of the ravages of excessive drink? Yet it does not follow that performing prenatal diagnosis of alcoholism is appropriate. If alcoholism is a genetic disease, then, presumably, genetic professionals will have expertise in its incidence, onset, course, and prognosis, and they will be familiar with appropriate professionals to whom persons with alcoholism may be referred for treatment. It is appropriate for the genetic professionals to bring all that information to bear in the genetic counseling process. It would be inappropriate for them to fail to do so because their professional expertise is what their patients are paying them for, and its use is the only justification for the existence of their profession. If at the time when this issue arises, alcoholism has become a readily treatable condition, the professionals should tell Mr. and Mrs. A that and should counsel them to avoid testing the fetus if the risks of stigmatization outweigh the burdens associated with the disease.

Mr. and Mrs. A will assert an autonomy interest in having their fetus tested. We normally remit medical decision making for children to the children’s parents, and at first blush there is no reason to refuse to do that before the child is born. Indeed, the case for parental, especially maternal, control is stronger before birth than after


10 See, sources cited op. cit. (fn. 6).
because of the need to involve the mother’s body in any diagnostic or treatment activity. Nonetheless, most commentators argue that presymptomatic diagnosis of children (and fetuses) for genetic diseases is inappropriate unless a medical intervention is possible if a positive diagnosis is made. I have argued against this position elsewhere, and I continue to believe that the substitution of professional for parental judgment about presymptomatic testing is usually an unjustified imposition of the inexpert personal views of persons who happen to be professionals.

In this case, no valid reason exists for refusing to test the fetus. Once the professionals have explained all the relevant information and engaged in directive counseling by advising the A’s not to have the fetus tested (if that is their professional opinion), then the A’s should remain free to make the ultimate decision. Otherwise, a refusal to test will reflect the professionals’ views about abortion, alcohol consumption and abuse, how significant and expensive treatments must be before they are too significant and expensive, etc. – all matters of social preference, not professional expertise.

The reason that the A’s judgment should prevail over the professionals’ is not that parental autonomy must always outweigh other values. It is that in this case the professionals have no more claim to wisdom than anybody else. Therefore, there is no reason to deviate from our ordinary preference for parental decision making. The normal reasons for that preference – parental wisdom exceeds the child’s; parents want what is best for their children; parental obligations to children should be accompanied by some parental rights – are valid in the absence of evidence that suggests something amiss in this case. Absent that, no justification for preferring the judgment of strangers exists.

Does this preference for parental decision making apply in the homosexuality context as well? Certainly, the effort by the fertility center to avoid the birth of a homosexual child is unacceptable. At worst it represents an imposition of prejudice. At best it is a paternalistic effort to spare the as yet unconceived child the negative effects of becoming a member of a stigmatized group. However, that cannot be justified here, even if one is open to paternalism in some contexts, because


12 Dworkin, 7 Ann. Rev. of Law and Ethics op.cit. (fn. 1).

13 Ibid., p. 28 and fn. 31 and sources cited therein. The argument that parental rights should accompany parental obligations is not strong in the case of selecting characteristics of a child before birth as the discussion of the homosexuality case will make plain. See, infra. However, the other arguments for deference to parents apply, and the absence of an additional argument is no reason to deviate from the ordinary state of affairs.

there is no professional basis for deciding that life as a homosexual is a net burden to a potential child. Imagine the outrage that would greet a suggestion that fertility services should only be made available to members of the dominant race to avoid creating a child who will be stigmatized and discriminated against.\(^1\) The problem here is the same.

It is harder to know how to respond to Mr. B’s and Mr. C’s efforts to guarantee the birth of a homosexual child. Those who are committed to parental autonomy will support their desire. Those, like me, who generally support parental control because it seems likely to work out best most of the time, will begin with a predisposition to do the same. Is this the best approach?

Parental autonomy is an inadequate basis for decision making. Every parental decision affects the unborn (here unconceived) child, and a decision to predestine the child to a particular style of life is a decision to deny autonomy to the child. Why should progenitors be free to insist that their offspring have certain characteristics?\(^16\)

The truth is that within limits parents do have the power to choose their children’s characteristics. Selecting a mate of one’s own race guarantees that the child will also be of the same race. Is it permissible to go farther than this “natural selection,” to enlist help in achieving a desired characteristic? For example, is it acceptable for a white couple that seeks artificial insemination by “donor” to insist that the sperm provider be white? If we assume (as I think most people would) that the answer is yes, then is it any less acceptable for a gay couple to insist that their child be gay?

First, it is entirely possible that the assumption that insisting on same-race artificial insemination is acceptable may turn out after analysis to be wrong. Even if it is correct, however, at least two differences between race and sexual orientation may be relevant. First, race is different than sexual orientation in that race is immediately apparent to a casual observer while sexual orientation (or the future sexual orientation of a child) is not.\(^17\) Thus, potential parents and their family-to-be have


\(^16\) One distinguished scholar has suggested that selecting offspring characteristics is a part of procreative liberty, John A. Robertson, “Genetic Selection of Offspring Characteristics,” in 76 B.U.L.Rev. 421 (1996), but there is an important difference between being allowed to procreate and being helped to do so, and there is another important difference between being allowed to procreate and being allowed to dictate the terms under which one will do so.

\(^17\) Berkowitz and Snyder make this point in the quite different context of arguing that parents should not be allowed to insist that their children be of a race different from the parents. They would insist upon, “reasonable phenotypic approximation.” Berkowitz and Snyder, op. cit. (fn. 15). pp. 35 – 37.
an extra interest in having a child who shares their race, an interest rooted in avoiding exposure to prejudice. Individuals who are not themselves racists are not obligated to volunteer to become martyrs in the war against racism. No extra prejudice or new prejudice against the parents or the family unit will be caused by gay parents having a heterosexual child.

Second, racial matching may be achieved almost effortlessly while sexual orientation matching requires genetic testing. Is that difference significant? I think it is. The fact that a person can achieve one questionable goal easily is no reason to facilitate his achieving other questionable goals that would require significant effort and outside assistance to achieve. The inability to stop a person from doing one thing does not suggest that we should facilitate that person's doing something similar. If assuring that their child will be gay is a deprivation of that child's autonomy, a commitment to B's and C's autonomy cannot justify the action. My autonomy to swing my fist ends where your nose begins.

But all sorts of parental decisions deprive parents' present and future offspring of autonomy. We usually accept that for the reasons noted above. Is there any reason not to follow the normal preference for parental decision making here? Normally, we assume that parents have a better sense of what is good for their child than the child does. Here, of course, the child does not exist, but B and C also have no claim to superior knowledge. This is not a case like a religious refusal of medical treatment case in which we are willing to defer to a parent's views of the relative weight of a child's health and the child's immortal soul. It seems hard to argue that the child will be better off with a minority sexual orientation. The parents are the ones who want this for their benefit, although they are likely to make an unpersuasive argument that the child will be reared better if it is exactly the way the parents want it to be. That argument, if taken seriously, would support almost any child-designing agenda that any parents might formulate.

Another reason to defer to parents is that they are presumed to care more about their child than anyone else does. Here there is no child to care about, and the parents' motivations seem selfish.

Finally, parents have many obligations to their children; we usually think that some rights should accompany those obligations. Here, however, as noted, there is no child. B and C have no obligations. The question is whether they have the right in advance to condition their obligations by dictating one of the potential child's characteristics. Clearly, the answer should be no. The ability to refrain from having a child does not imply a right to dictate the child's condition any more than a right to remove life support from a patient would imply a right to do something short of causing the patient's death, like beating him up. Moreover, parents' rights are rights to control some aspects of their children's upbringing (religion, schooling, etc.). They are not rights to have a certain kind of child. Parental obligations exist regardless of whether the child is the way the parents would like it to be. Parents who are disappointed by the birth of a child with a serious birth defect, may not
abandon the child because they did not get what they wanted. Similarly, parents should not be able to dictate what they want in advance.\textsuperscript{18}

Thus, the usual reasons to defer to parents do not exist. Given that, Mr. B and Mr. C should not be able to dictate the sexual orientation of their child for two reasons: First, there is a general presumption against designing children. This is based on recognition of human ignorance about what characteristics to promote and what undesired characteristics may accompany desirable ones and on a commitment to the value of genetic diversity. Second, the intentional creation of a child with a stigmatized characteristic that does not carry a clear compensating benefit is an act of cruelty to a child.\textsuperscript{19} This is not to say that it ought to be prohibited by the law. The suggestion is only that the request by B and C is unethical, and that the fertility center will be behaving ethically if it refuses to honor their request. To put the matter another way, being heterosexual is not a disease, and therefore parents ought not to insist that their children not be born with heterosexuality.

The same method of analysis can resolve the predisposition questions. Breast cancer and schizophrenia are clearly diseases, and they are diseases whose onset and course are less susceptible to control by the patient than alcoholism. Thus, if the certainty of the condition, rather than a predisposition to it were at issue, the parents would clearly be acting within ethical bounds to obtain prenatal diagnosis and, if they decided to do so, to terminate the pregnancy. Does the fact that only a predisposition exists change the answer?

In a sense, we are all predisposed to a fatal disease. When I use the term, "predisposition," to refer to the conditions of the fetuses here, I am assuming that it will be possible to determine that a particular fetus carries a mutation that significantly increases its risk, after birth, of contracting schizophrenia or breast cancer as compared to the background risk. Here, professionals understand the degrees of risk, the nature of the diseases and their treatments, and the current state of research for ultimate cures. Respect for their patients requires that they share that information and their opinions based on it with the potential parents. As long as there is no reason to doubt the parents' motives in wanting to spare their children from suffer-

\textsuperscript{18} Prospective parents are ethically permitted to try to avoid the birth of a child with a serious genetic disease or birth defect. This is to avoid suffering caused by disease and, often, to honor the values that underly a woman's right to choose to terminate her pregnancy. However, the opportunity to try to avoid the birth of a child with a birth defect does not imply either a right to abandon a child with a defect if one is born despite the effort to avoid it, or the right to select among nondisease states.

\textsuperscript{19} We need not resolve the question of whether being created with a stigmatized characteristic is a net benefit or detriment to the child who did not exist at the time the decision was being made. Only three situations are possible: No child will be born; a child with the stigmatized condition will be born; or a child without the stigmatized condition will be born. Less cruelty will occur if a child without a stigmatized condition is born than if a child with such a condition is born. If having a heterosexual child is unacceptable to B and C, they are, of course, free to refrain from having a child at all.
ing or the risk of suffering, the parents’ preferences should be followed even if the professionals disagree.

All of these cases suggest that focusing on the characteristics and limits of professionalism and on respect for relevant individuals leads to sound results in the genetic disease context. Unlike an autonomy-based analysis, this approach does not prevent us from evaluating and limiting parents’ conduct if the parents seek to control a child’s characteristics when the effect of doing so would be the prevention of a normal characteristic or the creation of a disease state. This analysis has implications for those who would suggest that deaf persons, dwarves, etc. have an entitlement to insist on offspring who share their conditions.

Zusammenfassung

Der Beitrag verwendet schwierige Fälle, um an ihnen die Angemessenheit einer Ethik der genetischen Beratung zu überprüfen, die auf die Ideen der Patientenautonomie und der Vermeidung von Bevormundung gegründet ist. Dabei stellt sich heraus, daß ein solcher Ansatz den Erwartungen, die an ihn zu richten sind, nicht gerecht wird. Es wird gezeigt, daß ein ethischer Ansatz, der auf Professionalismus und den Respekt vor dem Individuum setzt, eher geeignet ist, in realen Fällen zu befriedigenden Resultaten zu führen.

In diesem Beitrag wird insbesondere ein Fall analysiert, in dem ein Mann, dessen Gene ein Risikopotential für die Entwicklung von Huntington’s Disease aufweisen, eine Herztransplantation benötigt. Anhand des Falles wird die Angemessenheit von Ansätzen erörtert, die den Anspruch einer Person, die Huntington’s Disease entwickeln wird, auf eine Transplantation zurückweisen würden. Und es wird untersucht, inwieweit Genetiker dazu geeignet sind, die Legitimität von Gründen festzustellen, die Patienten haben mögen, wenn sie um eine präsymptomatische Diagnose nachsuchen. Dabei werden Fälle möglicher pränataler Diagnosen für Alkoholismus, Homosexualität und Prädispositionen für Brustkrebs und Schizophrenie erörtert, um auf diese Weise die Grenzen elterlicher Autonomie zu ermitteln.

Es zeigt sich dabei, daß eine Fokussierung auf Professionalismus und Respekt zu einer besseren Behandlung für die Patienten führt, zu mehr Interesse für das Wohlergehen von lebenden und zukünftigen Kindern und zu einem besseren Schutz für die realen Autonomie-Interessen der Patienten, als dieses die traditionelle Betonung von Patientenautonomie und Nicht-Bevormundung zu leisten vermag.