Ethical Considerations in Prenatal Diagnosis

ELENA A. GATES, MD, *San Francisco, California*

Prenatal diagnostic testing raises a number of important ethical issues, some related to diagnostic testing in general and others related to the special circumstances of pregnancy. These issues are most effectively addressed in the context of a broader understanding of the goals of prenatal diagnosis. Our dual obligations—to the pregnant woman and to the fetus—have an important influence on the goals of testing. Testing seldom leads to treatment beneficial to the fetus, but more often can be beneficial to the pregnant woman, particularly if the information provided enhances her ability to make sound decisions about reproductive matters. The process of prenatal diagnostic testing can, however, limit a woman's sense of control over the decisions made about her pregnancy. It can also provide an opportunity for third parties to become involved in what are usually considered private matters. It is therefore important that the process of testing include adequate counseling and follow-up and that the patient's confidence be respected. As prenatal diagnostic technology expands, both in terms of patients to be tested and diagnoses to be sought, society will face difficult questions concerning access to testing and the justification for its use.

(Gates EA: Ethical considerations in prenatal diagnosis, *In* Fetal Medicine [Special Issue]. West J Med 1993; 159:391-395)

Prenatal testing for the diagnosis of particular fetal conditions has become an important component of prenatal care for many women. Testing methods range from noninvasive technologies such as ultrasonography to invasive intrauterine techniques such as amniocentesis, chorionic villus sampling, and umbilical cord blood sampling. This testing raises a number of important ethical issues, including many common to diagnostic testing in general: the issues of informed consent, the appropriate indications for testing, privacy, confidentiality, and the just allocation of testing services and follow-up treatment.

Because this testing occurs within the context of pregnancy, important additional ethical considerations arise. The increasing availability of diagnostic and therapeutic procedures directed specifically toward improving fetal health has led some observers to consider the fetus to be a patient in its own right, a patient towards whom the obstetrician owes a duty of beneficence, even though the fetus resides within the body of a pregnant woman.[1-3] A good deal of controversy exists over the appropriate application of the concept of the "fetal patient" and includes discussions of the role of the pregnant woman as primary decision maker in matters regarding fetal well-being.[4-6]*

*See also the article, "Ethical Issues in Recommending and Offering Fetal Therapy," by F. A. Chervenak, MD, and L. B. McCullough, PhD, on pages 396-399.

"Autonomy is the moral right to choose and follow one's own plan of life and action."[7(p49)] Although the fetus is deserving of an obligation of beneficence on the part of the obstetrician, it is not an autonomous individual. It cannot be considered a patient in the same sense that the pregnant woman is.

Meaningful consideration of the ethical issues that arise in prenatal diagnostic testing requires a clear sense of the goals of the testing process. In the context of pregnancy, testing may be directed toward the maternal patient, the fetus, or both. The goals of testing will differ depending on which of these parties one posits as the possible beneficiary.

The Fetus

Diagnostic tests are generally used in medicine to benefit the person affected by the disease that a test reveals, while avoiding harm out of proportion to the benefit that the testing can provide. If a physician considering prenatal diagnostic testing regards the fetus as the intended beneficiary, this concept of the goals of testing becomes a problem.

Current methods of obtaining material for prenatal testing often involve risk to the fetus; the possible benefit to the fetus is less clear. The effective treatment of fetal

From the Department of Obstetrics, Gynecology and Reproductive Sciences, University of California, San Francisco, School of Medicine.

Reprint requests to Elena A. Gates, MD, Dept of Obstetrics, Gynecology and Reproductive Sciences, UCSF School of Medicine, Rm M-1495, San Francisco, CA 94143-0132.

conditions is certainly the ultimate aim of much of the technology developed in the area of prenatal diagnostic testing. Currently, however, actual benefit to the fetus or to the expected child can be achieved in only a few situations. When fetal anemia develops due to isoimmunization, prenatal therapy can be effective.[8] Physicians have had some success in treating immunodeficiency syndromes by fetal bone marrow transplantation.[9] The future child may also benefit if the condition diagnosed indicates a change in obstetric management. For example, if hemophilia is diagnosed in a fetus, scalp sampling and the use of a vacuum extractor to assist in delivery can be avoided for the protection of the newborn.

Unfortunately, prenatal testing has not yet led to meaningful therapy for fetuses affected with most of the conditions that can be identified. Even when treatment becomes possible before birth, difficult decisions about the relative risks of treatment, the effectiveness of the therapy, and the need for ongoing and potentially burdensome interventions will remain. One set of conditions for which prenatal diagnosis is frequently used, chromosomal aneuploidy, will probably never be ameliorated through fetal therapy. Selective abortion of the affected fetus will remain an important alternative.

This observation calls into question the appropriateness of much of what is available in terms of diagnostic testing and, particularly, invasive testing, if it is in fact intended to benefit the fetus. Currently used procedures generally involve risk, seldom lead to benefit for the fetus, and often result in a decision for selective abortion. The professional ethics of medicine prohibit the active killing of patients.[10,11] Physicians owe their patients a duty of nonmaleficence—an obligation to avoid doing harm. Whether sparing a fetus a future life of disease, pain, or disability is a benefit or even an avoidance of harm to that future child is a difficult ethical question on which a spectrum of articulate opinion exists.[12-14] If a postnatal life with Turner syndrome or even a more serious condition such as Down syndrome is considered a life worth living, it is difficult to conceive of selective abortion as a benefit or even an avoidance of harm for that potential child.[15]

The power to diagnose fetal conditions exceeds the power to treat them and probably will for a long time to come. Although future investigation may well lead to therapies for many genetic conditions and therefore be of benefit to fetuses and children, the conditions that are now most frequently diagnosed prenatally are ones that will, most likely, never be amenable to meaningful therapy. In these cases it is difficult to conceive of most testing as being beneficial to a fetus in any important way. Whereas the quest for fetal therapy may justify ongoing research in prenatal diagnosis, it does not justify the current expansion in the clinical use of this technology. Another goal for therapy would have to be postulated to support this practice.

The Pregnant Woman

Although the cells being analyzed belong to a fetus or fetal-placental unit, they are obtainable only by traversing the body of a pregnant woman. Whereas the condition being diagnosed is one affecting the fetus, the pregnant woman is the source for any decision that will be made in response to that diagnosis. Despite the fact that the pregnant woman does incur some risk, both physical and psychological, during the process of testing, she may benefit in terms of her ability to make reasoned choices about reproductive matters when test results are received.

Before a test is used in a clinical setting, physicians who intend to use it have an obligation to be certain that the test will provide results that are accurate and reliable. Furthermore, those recommending a diagnostic test must be confident that the test will, in general, yield the benefits that are its goal. If a pregnant woman is intended to be the beneficiary in most instances of prenatal diagnostic testing, and if the benefit expected consists of an enhancement of her ability to exercise informed reproductive choice, then it is important to assess whether that benefit is in fact being achieved. Making choices about pregnancy is a complex and personal process that takes place in the context of ongoing relationships with spouse, family members, and others. It involves the consideration of deeply held personal values and an assessment of one's life circumstances. The process of and response to prenatal diagnostic testing may therefore have an effect on many areas of a woman's life. The sum of these positive and negative effects will determine whether, overall, prenatal diagnostic testing is of benefit to her.

Reassurance

One way in which prenatal testing might benefit a pregnant woman would be by providing reassurance to her about the health of her fetus. Although testing—at least α-fetoprotein and chromosomal testing—tends to raise anxiety before the procedure and while test results are being anticipated, most women experience a decrease in anxiety levels to or below their baseline once normal results are received.[16-20] A group of women who received abnormally low results on α-fetoprotein screening were noted to be substantially more worried about their babies' health three weeks after testing—after normal follow-up results were available—than were women who received an initial normal result. These differences were not appreciable at later points in the pregnancy.[21] Dixson and co-workers observed women who received genetic counseling and then either underwent amniocentesis or declined.[17] The rate of continuing concern about possible congenital abnormalities was just over 20% in both the group who received normal amniocentesis results and in the group of women who declined testing.

The issue of reassurance can also be examined in the converse. Can the information that is obtained through prenatal diagnosis be falsely reassuring? Even if the specific diagnosis being sought, such as muscular dystrophy, is ruled out, approximately a 3% chance remains that the child will be born with some kind of congenital disorder or genetic disease. Will women who undergo prenatal genetic testing be more prepared or less prepared for other congenital problems that may arise?

Enhancing Autonomy

Eric Cassell, a physician and medical ethicist, has stated that "the . . . function of medicine is to preserve . . . autonomy."[22(p18)] This is consistent with the concept that an important goal of prenatal diagnostic testing is to enhance a pregnant woman's ability to make good choices about her pregnancy. Although it is true that prenatal diagnostic testing provides options to women at risk for bearing children with genetic diseases or other congenital conditions, it is not clear from existing data that reproductive choice is actually enhanced. Rapp has asked whether amniocentesis offers women "a 'window of control' or an anxiety-provoking responsibility."[23(p109)] Lippman notes that "some features of prenatal diagnosis do increase control but allocate it to someone other than a pregnant woman herself."[24(p34)]

Several concerns have been articulated regarding the influence of prenatal diagnosis on a woman's sense that the decisions made about her pregnancy are really her own. One survey of women undergoing amniocentesis or chorionic villus sampling indicated that 75% of them found it difficult to refrain from a prenatal diagnostic study once it was offered.[25] Of these women, 78% felt it would be more difficult to give birth to a disabled child if they had not accepted prenatal diagnosis. Many stated that whereas they felt "free from external pressure," they still felt an "obligation" to have testing done.

Among women of a certain age, a willingness to undergo prenatal diagnostic testing seems to be construed as a sign of responsible parenting. Is a woman acting irresponsibly in the eyes of others if the procedures are forgone? Might a woman sense less sympathy and support if testing is forgone and a child is born with a diagnosable condition or if a pregnancy is continued in the face of adverse results? If perceptions such as these are common among pregnant women, the availability of prenatal diagnosis may in fact limit autonomy rather than enhance reproductive choice.

Privacy

The availability of prenatal genetic testing has brought the interests and influence of family, friends, and society into a woman's pregnancy to an increasing degree. It has allowed third-party involvement in the most private of decisions being made during pregnancy. Involvement by parties such as employers and insurance companies in a woman's decisions about prenatal genetic testing could restrict reproductive choice in an unprecedented way.[26]

In one illustrative case, a pregnant woman whose living child was affected with cystic fibrosis sought prenatal testing for the disease. When testing revealed that the expected child would also be affected with cystic fibrosis, the woman and her husband faced an agonizing decision. They ultimately decided not to abort the fetus. Their health maintenance organization, which had originally approved the test, decided that it would not provide medical coverage for an affected child, a child who would be born with a "preexisting condition" because of the prenatal diagnostic procedure. As one journalist wrote, "The insurance company's message was clear: The parents could either abort the defective baby or struggle alone with the financial burden of a sick child" (L. Thompson, "The Price of Knowledge: Genetic Tests That Predict Dire Conditions Become a Two-Edged Sword." *The Washington Post,* October 10, 1989, WH7, col 1). Although the health maintenance organization ultimately capitulated and agreed to cover the child's treatment, its initial policy serves as a sign of the new limits on maternal choice that may evolve as the capacity for prenatal diagnosis expands.

Options

Given the current inability to effectively treat most conditions diagnosed in utero, many women who learn through prenatal testing that their fetus is affected have only two options: continue the pregnancy, anticipating and preparing for the birth of a child with expected handicaps, or terminate the pregnancy. Many women who undergo testing have not yet made a decision as to which option they will choose if testing indicates an abnormality.[17,25] Testing should not be contingent on a woman's willingness to undergo abortion. Although a substantial proportion of people do not consider abortion a morally acceptable alternative, others see it as an important reproductive option, particularly in the context of a prenatal diagnosis.

Although state legislatures in this country are planning and establishing programs to enable women, through genetic screening and prenatal diagnosis, to avoid conceiving or bearing children with serious congenital conditions, a trend toward limiting a woman's right to decide to abort an affected fetus is evident. It seems likely, in view of recent Supreme Court decisions, that state legislatures will have increasing power to define the circumstances under which abortion will be legally available.[27] Although many are currently optimistic that abortion will remain a legal option in this country, women are still faced with limitations on the accessibility of abortion services in terms of the number and geographical distribution of facilities offering abortion procedures. Women facing the termination of a desired pregnancy based on an abnormal outcome from prenatal testing may have even more difficulty because they must locate facilities that offer the termination of second-trimester pregnancies. If access to abortion is substantially restricted, one really must ask just what kind of choices prenatal diagnostic testing offers. How much benefit is knowledge in the absence of meaningful options?

If enhanced decision making on the part of pregnant women is the outcome that justifies using prenatal diagnostic testing, a generally invasive and somewhat risky technology, it is important to demonstrate that the technology actually achieves this end before testing is applied widely, not only to those women at increased risk for having an affected fetus, but to women in general. Investigation of the effect of prenatal testing on pregnant women and their ability to make thoughtful and uncoerced decisions about pregnancy should continue. Efforts to im-

prove the current testing process so that the benefits to pregnant women are increased will also be important.

Counseling

The importance of the nonclinical implications of test results, and the acknowledgement that a woman's personal values are of primary importance in the decision being made, are two of the considerations that underlie the concept of nondirective genetic counseling.[28] To enhance decision making by a patient, pretest counseling should provide accurate information about the testing procedure and the risks involved, including the possibility of ambiguous results. Both the medical and social consequences of the proposed tests and their results should be discussed. Counseling should also include a discussion of the particular condition or conditions for which testing is being done. A patient's informed consent for testing must then be obtained. All of this presumes a certain level of expertise on the part of the person providing the counseling. At present, it is unlikely that most women's health care professionals are as skilled as trained genetic counselors in either genetic knowledge or in the technique of counseling.[29] It may be difficult for many professionals to help patients identify personal preferences while avoiding the implicit communication of their own. There is evidence, for example, of an increased tendency for women to terminate a pregnancy affected by a sex chromosome abnormality when counseled by a general obstetrician rather than a geneticist.[30]

As the use of prenatal diagnostic testing expands, the current shortage of genetic counselors will be exacerbated.[31] Most women, particularly those who are offered testing as a routine part of prenatal care, will be counseled by their regular obstetric professional. These professionals are obligated to improve their skills so that their patients may benefit from the process of testing and so that the burdens that are a part of testing do not, on balance, result in harm.

Throughout the process of testing, it is essential that women feel assured that information shared with their health care professional will be held in confidence. Information obtained through prenatal testing should not be revealed to institutional third parties such as insurance carriers or employers without a woman's explicit consent.[32] The permissibility of sharing information about genetic diagnoses with other family members who are at risk, without the consent of the person being tested, is currently a matter for debate.[33,34] As a rule, patient confidentiality should be respected. A breach of confidentiality may be justified if the conditions set forward by the President's Commission for the Study of Ethical Problems in Medicine are met[35(p44)]:

- Reasonable efforts to elicit voluntary consent to disclosure have failed;
- A high probability exists that harm will occur if the information is withheld and the disclosed information will actually be used to avert harm;
- The harm that identifiable persons would suffer would be serious; and
- Appropriate precautions are taken to ensure that only the genetic information needed for diagnosis or treatment, or both, of the disease in question is disclosed.

Applying an Expanding Technology

In most circumstances, the process of prenatal diagnostic testing more effectively benefits a pregnant woman than it does the fetus actually being tested. Nevertheless, the obstetric professional still maintains important obligations to both parties. What effect does this dual obligation have when decisions are made about which conditions are reasonable targets for prenatal diagnosis?

Juengst has proposed that the range of appropriately diagnosed conditions be limited to those "relevant to the welfare of the fetus."[36] This approach, by providing information relative to the health of the fetus, can help parents make decisions in the context of fetal health. It also confines the process of prenatal diagnostic testing to the "usual context of medical practice," that is, health. Even within these limitations, however, decisions will be required as to the appropriateness of prenatal diagnosis and selective abortion for mild impairments such as von Willebrand's disease, treatable conditions such as phenylketonuria, late-onset conditions such as Huntington's disease and Alzheimer's disease, or the increased susceptibility to a condition such as coronary artery disease. As Botkin points out, "This technology will force us to examine the fundamental relationship between parent and child. How much control can we legitimately exert over the biologic structure of our children and how would such control change the way we view our children, how they view us, and how they view themselves?"[37(p104)] If a societal consensus on questions such as these cannot be reached, individual choice is likely to remain decisive.

Economic considerations may constrain our ability to provide prenatal diagnosis for conditions such as those just listed. Legitimate concern already exists about inequalities in access to what are currently considered routine prenatal diagnostic services. Many of these inequities reflect society's problems with access to prenatal care in general. If the affluent continue to have disproportionate access to technologies such as chorionic villus sampling and amniocentesis, being genetically disabled could become, as much as anything, a mark of social class.[34] This sort of inequity may increase as noninvasive screening technologies, such as the use of the "triple marker,"* become widely available.[38] A larger segment of society will have access to testing, with the exception of those with limited access to health care in general. As we face decisions about the wisest use of health care resources, a technology that provides options or enhances choice may not appear as essential to the public health as a technology that provides cures.

Conclusion

As perinatal medicine advances, more interventions will become possible, enhancing the potential benefit of

*See N. C. Rose, MD, and M. T. *Mennuti, MD*, "Maternal Serum Screening for Neural Tube Defects and Fetal Chromosome Abnormalities," on pages 312-317.

testing and expanding the treatment options available for affected fetuses. A substantial proportion of testing, however, will continue to be done for conditions such as Down syndrome for which meaningful prenatal treatment, and therefore fetal benefit, is not expected. In these contexts, the pregnant woman, often in conjunction with her partner, will remain the principal beneficiary of the process of prenatal diagnosis. As individual decisions are made about the use of prenatal diagnostic technology, it will be important that obstetric professionals be able to identify the goals of the particular test being considered, in relation to both a pregnant woman and her fetus. Health care professionals will have an important role in helping pregnant women and their partners elicit their own preferences about testing and come to well-considered decisions in response to testing results.

REFERENCES

1. Harrison MR, Golbus MS, Filly RA (Eds): The Unborn Patient. Philadelphia, Pa, WB Saunders, 1990

2. Cunningham FG, MacDonald PC, Leveno KJ, Gant NF, Gilstrap LC (Eds): Williams Obstetrics, 19th edition. Norwalk, Conn, Appleton & Lange, 1993

3. Chervenak F, McCullough LB: Perinatal ethics: A practical method of analysis of obligations to mother and fetus. Obstet Gynecol 1985; 66:42-47

4. Ruddick W, Wilcox W: Operating on the fetus. Hastings Cent Rep 1982; 12:10-14

5. Fletcher JC: The fetus as patient: Ethical issues. JAMA 1981; 246:772-773

6. Committee on Ethics—Patient Choice: Maternal-Fetal Conflict. Washington, DC, American College of Obstetricians and Gynecologists, 1987

7. Jonsen AR, Siegler M, Winslad W: Clinical Ethics. New York, NY, Macmillan, 1986

8. Berkowitz RL, Chitkara U, Goldberg JD, Wilkins I, Chervenak FA, Lynch L: Intrauterine intravascular transfusions for severe red blood cell isoimmunization: Ultrasound-guided percutaneous approach. Am J Obstet Gynecol 1986; 155:574-579

9. Touraine JL: Rationale and results of in utero transplants of stem cells in humans. Bone Marrow Transplant 1992; 10[suppl]:121-126

10. Brock D: Voluntary active euthanasia. Hastings Cent Rep 1992; 22:10-22

11. Wanzer SH, Federman DD, Adelstein SJ, et al: The physician's responsibility toward hopelessly ill patients: A second look. N Engl J Med 1989; 320:844-849

12. Liu AN: Wrongful life: Some of the problems. J Med Ethics 1987; 13:69-73

13. Botkin JR: The legal concept of wrongful life. JAMA 1988; 259:1541-1545

14. Elias S, Annas GJ: Reproductive Genetics and the Law. Chicago, Ill, Year Book Medical Publishers, 1987, pp 113-120

15. Fost N: Guiding principles for prenatal diagnosis. Prenat Diagn 1989; 9:335-337

16. Beeson D, Golbus MS: Anxiety engendered by amniocentesis. Birth Defects 1979; 15:191-197

17. Dixson B, Richards TL, Reinsch RN, Edrich BS, Matson MR, Jones OW: Mid-trimester amniocentesis: Subjective maternal responses. J Reprod Med 1981; 26:10-16

18. Evers-Kiebooms G, Swerts A, van den Berghe H: Psychological aspects of amniocentesis: Anxiety feelings in three different risk groups. Clin Genet 1988; 33:196-206

19. Finley SC, Varner PD, Vinson PC, Finley WH: Participants' reaction to amniocentesis and prenatal genetic studies. JAMA 1977; 238:2377-2381

20. Robinson GE, Garner DM, Olmsted MP, Shime J, Hutton EM, Crawford BM: Anxiety reduction after chorionic villus sampling and genetic amniocentesis. Am J Obstet Gynecol 1988; 159:953-956

21. Marteau TM, Cook R, Kidd J, et al: The psychological effects of false-positive results in prenatal screening for fetal abnormality: A prospective study. Prenat Diagn 1992; 12:205-214

22. Cassell E: The function of medicine. Hastings Cent Rep 1977; 7:16-19

23. Rapp R: Moral pioneers: Women, men and fetuses on a frontier of reproductive technology. Women Health 1987; 13:101-116

24. Lippman A: Prenatal genetic testing and screening: Constructing needs and reinforcing inequities. Am J Law Med 1991; 17:15-50

25. Sjogren B, Uddenberg N: Decision making during the prenatal diagnostic procedure. A questionnaire and interview study of 211 women participating in prenatal diagnosis. Prenat Diagn 1988; 8:263-273

26. Gates EA: Maternal choice: Will it work both ways? Womens Health Issues 1990; 1:25-27

27. *Planned Parenthood v Casey*, 91-744, Slip Op (1992)

28. Hsia YE, Hirschhorn K: What is genetic counseling? *In* Hsia YE, Hirschhorn K, Silverberg RL, Godmilow L (Eds): Counseling in Genetics. New York, NY, Alan R. Liss, 1979, pp 1-29

29. Kershner MA, Hammond EA, Donnenfeld AE: Knowledge of genetics among residents in obstetrics and gynecology (Abstr). Am J Hum Genet 1992; 51(S):A16

30. Holmes-Seidel M, Ryynanen M, Lindenbaum RH: Parental decisions regarding termination of pregnancy following prenatal detection of sex chromosome anomalies. Prenat Diagn 1987; 7:239-244

31. Meyer JL, Edwards JG, Young SR, Brooks KA: Human resource study on genetic counselors (Abstr). Am J Hum Genet 1992; 51(S):A269

32. Wertz DC, Fletcher JC, Mulvihill JJ: Medical geneticists confront ethical dilemmas: Cross-cultural comparisons among 18 nations. Am J Hum Genet 1990; 46:1200-1213

33. Fost N: Ethical issues in genetics. Pediatr Clin North Am 1992; 39:79-89

34. Wertz DC, Fletcher JC: Ethical issues in prenatal diagnosis. Pediatr Ann 1989; 18:739-749

35. President's Commission for the Study of Ethical Problems in Medicine and Biomedical and Behavioral Research: Screening and Counseling for Genetic Conditions. US Government Printing Office, 1983

36. Juengst ET: Prenatal diagnosis and the ethics of uncertainty, *In* Monagle JF (Ed): Medical Ethics: A Guide for Health Professionals. Rockville, Md, Aspen Publishers, 1988, pp 12-25

37. Botkin JR: Ethical issues in human genetic technology. Pediatrics 1990; 17:100-107

38. Phillips OP, Elias S, Shulman LP, Andersen RN, Morgan CD, Simpson JL: Maternal serum screening for fetal Down syndrome in women less than 35 years of age using α-fetoprotein, hCG, and unconjugated estriol: A prospective 2-year study. Obstet Gynecol 1992; 80:353-358