

Public Policy Issues Raised by a Medical Breakthrough

AMITAI ETZIONI

Amniocentesis, which detects mongolism and other serious abnormalities in a fetus in utero, will prevent the birth of thousands of afflicted children yearly once current research on its safety justifies its wide use. But this genetic intervention raises crucial questions of public policy, and lest we risk doing to our bodies what we have done to the environment, we must face them now. For example: Who shall be tested—all women who want to be or only those in high-risk categories? What shall the test be used for—diagnosis of debilitating diseases only, or also of the XYY (“criminal”) chromosomes, or even for “breeding purposes”? And whatever the question, who shall decide the answer?

According to two recently published studies, about half of all children born afflicted with mongolism need not be. Dr. Sarah Bunday, reporting in the September 3, 1973 issue of *Modern Medicine*, writes: “If all mothers 35 years of age and older were screened during pregnancy and therapeutic abortion done in every instance of trisomy-21 [the technical indication of Mongolism], the incidence of Mongolism would be halved.” While this finding is based on data from Great Britain, a report by Drs. Zena Stein, Mervyn Susser, and Andrea Guterman in the February 10, 1973 issue of *Lancet*, using American data, reaches a similar conclusion. Yet over the next twelve months roughly 14,000 mongoloid children will be born in the United States alone.

Dr. Aubrey Milunsky of Harvard has estimated that each year over 20,000 infants in the U.S., and over 700,000 worldwide, will be born with either mongolism or one of the other serious chromosomal ab-

normalities that can now be detected and averted.¹ Many of these children will die before they reach the age of seven, heart troubles and leukemia being the most common causes of death. The rest will suffer from varying degrees of mental retardation as well as from complications due to the malformation of one or more of their vital organs. Many, ignored by their parents and removed from society's view, will end up in public institutions which are often the contemporary equivalent of the notorious nineteenth-century "snake pits." The estimated cost to the American public of this primarily custodial and ameliorative, not curative, care is reaching \$1.7 billion a year.

The medical procedure which, if more widely utilized, could alleviate much of this suffering and cost is amniocentesis. The technique entails withdrawal of a sample of the fluid in which the fetus floats, somewhere between the fourteenth and eighteenth weeks of pregnancy. The fetal cells found in the fluid are then cultured and studied for chromosomal abnormalities. Illnesses other than mongolism can also be detected: these include serious sexual abnormalities (Turner and Klinefelter syndromes) and galactosemia (which can cause cataracts, cirrhosis of the liver, and mental retardation). If the test is "positive" and intervention is desired, abortion is necessary. Because of the "lateness" of the abortion, often toward the end of the fourth month of pregnancy or beyond, the use of hypertonic saline is considered the safest way to terminate the pregnancy.

While any woman who desires amniocentesis can approach a qualified physician and ask for the test to be performed, at present the procedure is not widely used; many women are unaware of it and many physicians will not recommend it. In a survey just completed by sociologist Nancy Castleman at the Center for Policy Research,² gynecologists chosen via a national random sample were asked whether or not they would recommend the procedure. A mere 4.7 percent answered in the affirmative; 90.2 percent in the negative. Asked whether they would recommend it for women over 40, the age at which the risk of mongolism rises sharply (from 1 in 600 for all women to 1 in 100 for those above 40), 59 percent indicated that they would, while 31 percent indicated that they would not.

1. Aubrey Milunsky, *The Prenatal Diagnosis of Hereditary Disorders* (Springfield, Ill.: Charles C. Thomas, 1973).

2. Nancy Castleman and Amitai Etzioni, "Amniocentesis: A Forerunner of the Genetic Fix," *Connecticut State Medical Journal* 38 (September 1974).

The reasons why some physicians will not recommend amniocentesis even for older women are many, ranging from religious opposition, to the abortion entailed if the test findings are to be acted upon, to simple resistance to a new procedure. The most important reason, though, is caution. Thus far, amniocentesis has been used in only several hundred cases, and the risks it poses to mother and fetus are still being evaluated. In some instances it may cause miscarriage, an infection in the mother, or—in very rare cases—damage to the fetus.

In order to assess the safety and effectiveness of this new medical procedure, an unusually intensive research effort is being undertaken: nine medical centers—among them Yale in New Haven, Mount Sinai in New York, and the UCLA and San Diego Medical Schools—are collaborating in a study comparing a thousand amniocentesis test subjects to a matched control group of 1,000 pregnant women not given the test. The study is being coordinated by Dr. Charles Lowe, the scientific director of the National Institute for Child Health Development, Bethesda, Md. While I was allowed to sit in on a recent day-long meeting of the study directors, I am committed, like all of them, not to comment on the partial findings of the study until it is completed. Final results summarizing the effects of the test on the mothers will be available in about a year, and data on its effects on the infants, somewhat later.

However, public debate and policymaking on several of the critical issues raised by amniocentesis must not be delayed until after the findings of the super-study are compiled. Fortunately, such an assessment is possible now. In the first place, the results of studies using smaller numbers of patients than in the super-study are available. Second, unlike drugs which are not permitted on the market until *after* they have been tested, new medical procedures remain on the market until they are found unsound. While amniocentesis is being evaluated, doctors are continuing to provide it to patients other than those included in the study population. Third, the issues the public and public authorities will have to face once the procedure is fully evaluated are highly complex; we would be wise to reflect upon them now and thus be armed with some forethought when the time for decision arrives. Most important, as we shall see below, the main policy questions raised by amniocentesis are predominantly normative, dependent only in part on medical considerations. They need not await the results of the medical study.

THE ISSUES

There are four complex normative issues of concern: who shall be tested, what shall the test be used for, what are the costs and benefits of the test, and how can patients in need of the service best be reached?

Who Shall Be Tested? Because the procedure itself entails a measure of risk, the most elementary question to ask is whether we should test every pregnant woman or only those who have a high risk of bearing an afflicted child. Because there is a clear association between incidence of mongolism and age of mother, as we reported above, few doctors favor testing every pregnant woman. The majority favor limiting the test to older women, 40 being the most agreed-upon age at which to start. On the face of it, this appears to be a strictly medical issue: given the fact that the test itself entails a small, but not trivial, risk, it should be given only to those who have a high likelihood of benefiting from it. Actually, the issue is highly charged with personal and moral values. The question each woman, her husband, and their physician must ponder is *how* strongly the couple feels about the possibility of having a mongoloid child—some parents feel that this would be a catastrophic event, while others feel that they could learn to accept it—compared to how undesirable a test-induced miscarriage (or other complication) might be. Similarly, the same test-associated risks would probably seem quite different to a woman who had already borne several children, and was considering an abortion anyway, than to an older woman pregnant for the first time. The answer which suggests itself is that the decision should be made by each woman (or couple) *after* they have been thoroughly *informed* about the risks involved in the test—and in not testing.

What Shall the Test Be Used For? Aside from revealing some severe illnesses, as already indicated, the test also detects milder illnesses as well as genetic attributes which may be asocial but not a danger to health (e.g., the so-called “criminal chromosomes” XYY). Finally, it indicates sheer physical attributes (the sex of the fetus).

Writing in the August 1973 issue of *Prism*, Dr. Albert Dorfman, Director of the Joseph P. Kennedy Jr. Mental Retardation Research Center, draws a distinction between severely debilitating diseases entailing limited life expectancy and less serious illnesses, such as Farby's, with which the child suffers severe physical problems but as a rule survives and grows up without mental retardation. Dr. Dorfman

goes on to voice his concern, shared by other doctors, over whether it is reasonable to carry out prenatal diagnosis and abortion of fetuses afflicted with other, even milder and treatable, illnesses (such as diabetes) which may soon be detectable via amniocentesis. In the quest for ever more "perfect" babies, there must be a place where the risks and costs of testing and abortion outweigh the gains. But where should the line be drawn, and by whom?

If that question is a tough one, ponder the next: What should be done about a fetus which has XYY chromosomes? True, available data on the effects of the XYY chromosomes are inconclusive and highly controversial. Nevertheless, there is *some* evidence that persons with XYY have a somewhat higher chance to end up criminally insane. Should parents be told when the test shows the fetus to have an XYY profile? If they are told, will not thousands of otherwise normal fetuses be aborted, including a high percentage of those with XYY who would *not* have become criminally insane? If the parents are not to be told, on what grounds can such withholding of information be justified? Should the public interest enter the picture? In view of our great concern with crime, is it in our interest to promote such a screening?

As for the determination of desirable physical traits, there are instances on record where the test has been used by parents as a basis for deciding to abort a male (or female) fetus because they wanted a child of the other sex. In a survey just completed by Dr. James Sorenson of Princeton,³ 96 percent of the genetic counselors questioned opposed such a use of amniocentesis, the main reason being that it entails taking a medical risk for a non-health purpose. Still, if parents have the patience to shop around long enough, they can find a doctor who will administer the test for sex choice. Should amniocentesis be outlawed for such "breeding" purposes? And if so, on what grounds, now that abortion is legal "on demand"? Or from the viewpoint of the public interest, should laboratories be limited to illness tests *until* enough testing capacity is available for illness-determination and only then be allowed to test for sex?

Cost-Benefit Analysis—We have learned to worry about the costs of new interventions lest they exceed the benefits. After all, hundreds of women must be tested before one afflicted fetus will be found. Dr.

3. James Sorenson, *Social Aspects of Applied Human Genetics* (New York: Russell Sage Foundation, 1971).

Aubrey Milunsky⁴ has estimated that it would cost about \$60 million a year to test all pregnant women aged 35 or older (each test costs about \$150). Assuming that all women told they were carrying genetically afflicted fetuses were to seek abortions, the added costs would be \$3,250,400. But the cost of institutional care for these children, if born, is conservatively estimated at \$460 million *a year*—and at about \$2 billion over their lifetimes. This burden must often be shouldered by the taxpayers. Is it “worth it,” then, for society to encourage the use of amniocentesis? Is it in the public interest to make the test cheaper? Free? At least for persons who cannot afford it?

Public Education Campaigns—The Nixon administration (and the Ford, it seems), I am told by civil servants who refuse to be cited, had a strong desire not to be associated with any programs which entailed or implied abortion (or, for that matter, even birth control). A brochure entitled “Mongolism, Hope through Research,” published by the Department of Health, Education, and Welfare, carefully plays down amniocentesis and does not mention at all that it may involve abortion. And a new brochure about amniocentesis, recently issued by HEW, avoids discussion of abortion. Should we not begin designing a major and detailed educational campaign now, so that when the super-study findings are available the public can be properly alerted to their implications?

WHO SHALL DECIDE?

The foregoing policy issues and others surrounding amniocentesis are not unique to this particular medical procedure. They are also raised by other recent developments in the area where genetics and medicine meet, such as mass screening for sickle cell anemia and premarital gene testing. Overarching all the specific issues relating to specific medical techniques is the question of who should make these complex and literally vital decisions. The more personal decisions—whether or not to have the testing done, whether or not to proceed with abortion—can be left to pregnant women and their husbands, in consultation with their physicians. But who shall pass on the whole range of public policy issues involved? Who shall decide whether the government should subsidize the costs of the amniocentesis test, or whether a crash program to expand lab facilities should be launched,

4. Milunsky, *Prenatal Diagnosis of Hereditary Disorders*.

or whether a major public education campaign should be undertaken? Should Congress make some decisions, passing a law forbidding the use of amniocentesis for sex choice, or forbidding its use without the husband's consent?

The technical questions could be dealt with easily enough by appropriate agencies; for example, issues of public education could be decided by the Surgeon General, and those of lab development by, say, the Public Health Service (prodded into action—I hope—by the respective congressional committees). But the main public policy issues require a higher-level, more encompassing and coordinated review than the crazy quilt of federal agencies and congressional committees can provide. Senator Walter Mondale's bio-ethics bill seems to me to provide the needed mechanism for such a comprehensive overview. Senator Mondale suggests that a congressional bio-ethics commission be set up to deliberate on these matters and to formulate appropriate guidelines. Such a commission would be composed of fifteen professionals in fields ranging from law to medicine, theology to technology. It would be granted a research staff and an annual budget of one million dollars.

Some might argue that such a commission is no longer needed, as Congress has recently passed Senator Edward Kennedy's bill providing for a commission to regulate experiments with human subjects. However, little if any duplication is involved. The Kennedy Commission is regulatory in intent and sharply focuses on one significant but narrow issue: the regulation of research lest scientists abuse those mental patients, prison inmates, or fetuses who serve as experimental subjects. The Mondale Commission would be reflective in nature and much broader in scope, covering not only those issues raised by amniocentesis (which concern every pregnant woman, husband, *and* tax payer) but also the myriad questions raised by other recent breakthroughs in medicine, from organ transplants to the right to die with dignity, from mass genetic screening programs to test-tube babies. Whether these deliberations would take place in a congressional commission, as Mondale suggests, or in a presidential one, or via some other societal-guidance mechanism, their purpose would be to answer our need for a systematic and comprehensive perspective on new genetic interventions. Of these, amniocentesis is but a forerunner; there are many more to come.

A GENETIC "STEAM ENGINE"

We are like the citizens of Britain when they first saw the steam engine. Few perceived, and even fewer acted upon, the notion that this technological innovation, in conjunction with others soon to follow, would promote very far-reaching societal changes—changes whose political, economic, and cultural consequences would amount to what is now known as the Industrial Revolution. Amniocentesis and mass genetic screening are but the path-breakers of ever more far-reaching genetic interventions which even now are being readied. Down the road are artificial wombs, cloning (making genetic copies of persons via asexual reproduction), and genetic surgery. More and more genetic procedures are turning issues heretofore settled by nature into matters which we must decide, both as individuals and as members of society. We must pay greater attention to the normative and policy questions that these new tools pose for us, so that we can channel their consequences more effectively than we did those of the Industrial Revolution, when we allowed technology to adapt society to its logic and needs. Our tardy treatment of the issues raised by the new breakthroughs suggests that we have learned little from our past. Once again we are proceeding in an ad-hoc manner. Will we delay in our examination of where bio-medical technology is leading, thus doing to our bodies what we have already done to our environment? Will we again limit our policy to corrective measures, after the damage is done? Or this time will we reflect upon the kind of future we want, anticipate alternative courses of action, and choose among them—before the genetic Pandora's box is fully opened?