Implications of Findings From the Amniocentesis Registry for Public Policy

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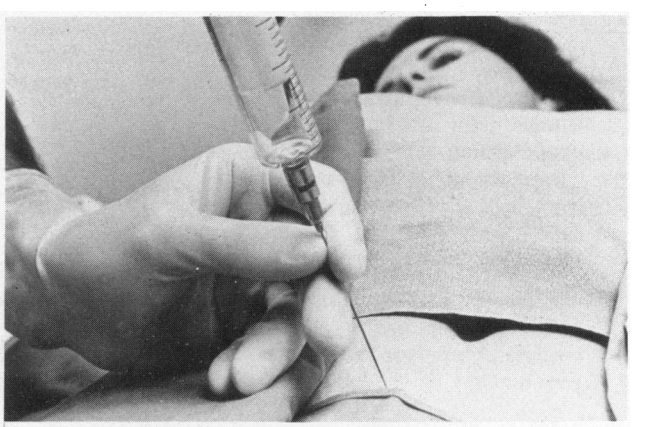
THE NATIONAL AMNIOCENTESIS REGISTRY study was conducted between 1971 and 1975 at nine medical centers under contracts from the National Institute of Child Health and Human Development. Information was obtained on immediate complications of the procedure, pregnancy outcome, and status of the child at birth and at 1 year of age for 1,040 women undergoing amniocentesis and for 992 matched control patients who did not. The incidence of any minor complication of the procedure (that is, vaginal bleeding, amniotic fluid leakage) was approximately 2 percent. There was no significant difference in the incidence of fetal loss between the amniocentesis group (3.5 percent) and the control group (3.2 percent), in prematurity, or in complications of labor and delivery, although caesarian section was performed more frequently in the amniocentesis group. On physical examination of the newborns, there was no significant difference between the two groups of infants in the incidence of congenital anomalies or complications after delivery. The two groups of infants were re-examined at approximately 1 year of age, and there was no difference in incidence of abnormal physical findings, number of illnesses in the first year of life, physical growth, or neurological and mental development.

Of the 1,040 women receiving amniocentesis, 45 (4.3 percent) were found to be carrying an abnormal fetus; 35 of these women and 4 others elected abortion. There were 6 erroneous diagnoses made for the 1,040 women, an accuracy rate of 99.4 percent. It was thus concluded that midtrimester amniocentesis is both an extremely safe and highly accurate diagnostic procedure for the detection of genetic disease.

These research results represent, in my opinion, fulfillment of the clinical research mission of the Public Health Service at its best. A need was perceived for prospective evaluaton of a new clinical procedure, a study was scientifically designed to provide the answers needed, and Public Health Service resources were targeted to obtain the information as quickly and economically as possible. As a result we can now say with virtual certainty that midtrimester amniocentesis for prenatal diagnosis is a safe technique that can be applied to larger segments of the population without undue risk or hazard. Amniocentesis thus moves clearly from the realm of a research procedure to a part of clinical practice. And we have this answer only 7 years after use of the procedure was first described. Would that we could be in the same position with all new medical procedures and techniques.

But research is only one part of the mission of the Public Health Service. We have clear responsibilities in public health education and in assuring availability to the public of the best in preventive health services as well. Medicine is often criticized for the lag between development of an advance and the time it becomes generally available to the public. Let this not be the case with amniocentesis. We do not have the excuse in this instance that the safety and accuracy of the techniques have not been established. We have those answers. Let us proceed to make the best use of them.

One of the main points of emphasis in the Department of Health, Education, and Welfare is prevention of disability. By focusing on prevention, we increase the resources available for other programs. Few advances compare with amniocentesis in their capability for prevention of disability. If we consider economic factors



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alone, and look only at prevention of Down's syndrome, the benefit to cost ratio for amniocentesis is well in excess of 10 to 1. But human considerations are even more important. With this technique we can now assure the older woman who is pregnant that she need not fear birth of a child with Down's syndrome and a consequent lifetime devoted to the care of a handicapped child. We can assure couples who are carriers of Tay-Sachs disease that they can have normal children without facing the agonizing process of helplessly watching an affected child slowly die. For these reasons it is most appropriate for the Public Health Service, as a matter of policy, to foster use of amniocentesis by those women for whom it is indicated by educating both physicians and the public as to the availability and applicability of the technique and, based on results of this study, its safety. The National Institute of Child Health and Human Development has begun a modest effort in this direction, targeted to reducing the incidence of Down's syndrome and one that is somewhat limited in part because of hesitation pending final results of this study. With the announcement of these results, there is no longer reason for hesitation. To the initial efforts by National Institute of Child Health and Human Development should be added educational efforts from all parts of the Department, that can reach the entire population. Physicians and women must know about amniocentesis, its indications and its potential benefits, so that pregnant women who wish to do so can avail themselves of this procedure.

Although advocating amniocentesis when indicated, the Department recognizes that the decision must always be that of the pregnant woman. We shall not imply coercion in any of our efforts, regardless of the risk of the pregnancy or the degree of dependence of the woman on the public for her medical care.

Publications of the results of this study, combined with the intended educational initiative of the Public Health Service, will assuredly result in an increased demand for amniocentesis services. Physicians will need to be trained in proper techniques for performing the procedure. The medical profession is capable of responding to this need without prodding or interference from the government. What we must do is assure that financial barriers to access to this procedure do not exist. Because of both the human considerations and the financial advantages of prevention of disability, we shall attempt to assure coverage for this procedure by both the private and the public medical insurance programs.

But the limiting factor in expanding amniocentesis is not likely to be physicians so much as laboratory facilities. Many of the laboratory analyses of amniotic fluids currently done in this country are performed in research laboratories. The work is thus supported by research grants. Some laboratories charge for the analysis on a sliding fee scale; others provide the service free. As amniocentesis passes from a research technique to accepted clinical practice, using research funds to support amniotic fluid analyses will become less and less attractive, if not impossible. What is the best method to provide laboratories that will do these analyses when such work has lost the excitement of research and become routine, while at the same time providing the quality assurance required by the life-death decisions hanging on the test results? This concern becomes of particular importance considering that even in the best of hands, as in this study, some errors may occur. Because of the high costs involved in gearing up a nationwide analysis capability, government involvement is probably necessary and appropriate.

Accordingly, I have begun discussions with officials in the Public Health Service, which are continuing, as to how best to meet this need. One approach which seems the most attractive would be for the Public Health Service to begin to establish incrementally a network of State or regional laboratories to perform cytogenetic analyses on amniotic fluid samples. Only a relatively small number of large laboratories would be needed. Since the increased demand will be almost entirely for cytogenetic analyses, these regional laboratories would perform only these studies; existing research laboratories would, for the near future at least, continue to provide the facilities for biochemical analyses, with a mechanism to minimize duplication of tests but with assurance that each test needed is available somewhere.

Ideally, these initial regional laboratories would be established under a contract at preexisting university centers, to take advantage of existing equipment and personnel, and under the direction of an experienced investigator to assure quality control. The government would provide seed money for space, equipment, and training additional personnel, with the eventual goal being a self-sustaining laboratory that would be supported by the fees charged for analysis.

Another alternative under consideration would be to establish a nationwide analysis capability under the aegis of the Center for Disease Control, working with State and local institutions as we do with other kinds of testing. In fact, a pilot project, in collaboration with a State health department and university, is already underway. Whatever method is chosen, we will put forth our efforts to assure that the laboratory capability exists to take advantage of this new advance in preventive medicine.

Our research efforts will not stop, however. There are enough new worlds left to conquer in prenatal diagnosis and therapy that gradual shifting of laboratory focus from cytogenetic analyses of amniotic fluids to related areas need pose no threat to the innovative investigator. Many other birth defects remain undetectable. Furthermore, a preventive technique dependent on elective abortion is not a final answer to the problem of birth defects. The progress reported at this symposium is but one more step along the long road to conquest of the disorders. But it is a step well worth taking, and one to which the Public Health Service will be responsive.