Am. J. Hum. Genet. 47:899–903, 1990

Maternal Serum Alpha-Fetoprotein Screening Activities of State Health Agencies: A Survey

George C. Cunningham and Kenneth W. Kizer

Genetic Disease Branch, California Department of Health Services, Berkeley

Summary

Maternal serum alpha-fetoprotein (MSAFP) screening has been demonstrated to be cost-effective on a population basis and is becoming standard practice. The American Society of Human Genetics has twice published policy statements to define the essential elements of a quality screening program. The present study reviewed the impact of these policy statements on state public-health agencies with respect to regulation or provision of MSAFP screening in their jurisdictions. With a few exceptions, states have not elected to play a major role in provision or regulation of this test. There is need to address issues of funding, standards, and data collection in a collaborative effort, if policy statements on genetic services are to be translated into effective state population screening.

Introduction

Maternal serum alpha-fetoprotein (MSAFP) screening for neural tube defects and other birth defects began in the United Kingdom in the late 1970s. In 1983, after extensive delays and public debate, the U.S. Food and Drug Administration adopted regulations approving several alpha-fetoprotein (AFP) testing kits, making expanded use of the test possible in the United States. Since then, MSAFP screening has proved to be a valuable and cost-effective means of improving pregnancy outcome (Main and Mennuti 1986; Tosi et al. 1987; Wald and Cuckle 1987).

On November 2, 1986, The American Society of Human Genetics (ASHG) developed a policy statement detailing the conditions necessary to provide for appropriate use of this test. This policy statement was sent to the chief administrative officer (director or commissioner) of each state health agency, to the nine regional genetic network directors, and to federal Maternal and Child Health (MCH) offices. Copies were provided to many professional groups, including the American Public Health Association. The statement was published in the February 1987 issue of *The American Journal of Human Genetics* (The American Society of Human Genetics 1987) and has been reproduced in other journals. In April 1989 the ASHG policy statement was updated and again similarly distributed (Garver 1989).

In view of the increasing interest in and acceptance of MSAFP testing by the medical community and the public, a survey was conducted in August 1987 to determine the extent to which decision makers in public health were aware of the ASHG guidelines and how the public-health community had addressed this issue. The same survey was repeated in January 1990 to determine changes in response. The present report discusses the results of these two surveys.

Methods

In 1987 the survey instrument consisted of eight questions. Two questions were added in 1990. The 1987 questionnaire asked (1) whether there were state laws or regulations pertaining to MSAFP, (2) whether there were state health laboratory activities in this area, (3 and 4) where testing was available, (5) the percentage of pregnancies being tested, (6) whether the respondent was aware of the ASHG policy statement, (7) what efforts had been made to translate the statement into public policy, and (8) which organizational unit was responsible for AFP testing. In 1990 we also asked what

Received October 13, 1988; final revision received July 30, 1990.

Address for correspondence and reprints: George C. Cunningham, M.D., Chief, Genetic Disease Branch, 2151 Berkeley Way, Annex 4, Berkeley, CA 94704.

0002-9297/90/4706-0004$02.00

priority had been assigned to MSAFP testing—and about the limitations to MSAFP programming—from the respondent's perspective. The questionnaires were sent to all state health officers and to all state MCH and public-health laboratory directors. Written or telephone responses were received from all states in both surveys.

Results

In 1987 only four states (California, New York, Maryland, and Iowa) had adopted laws addressing MSAFP screening. All of these states were aware of and attempting to follow the guidelines. In California, the Department of Health Services contracts for testing in eight state-approved and -monitored laboratories and provides complete follow-up services (i.e., counseling, ultrasound, and amniocentesis) in a fee-supported, statewide program administered by the state. This unique program, which began in April 1986, follows all ASHG recommendations.

New York limits testing to laboratories meeting state standards. Regulations define qualifications of laboratory directors, staff standards for quality control, and record-keeping requirements. On-site inspections are made, and laboratories are proficiency tested. There is no organized control of follow-up procedures, such as counseling and ultrasound, but amniotic fluid analysis for AFP and acetylcholinesterase are also regulated. The state health department provides blood tests for some recipients of publicly funded medical care. In 1987 New York had approved 13 in-state and three out-of-state laboratories for MSAFP testing. In 1990 the number of in-state laboratories is unchanged, but there are now eight out-of-state laboratories approved for MSAFP testing.

Maryland has regulations very similar to those of New York but also addresses issues of medical practice, such as requiring patient education and follow-up procedures. The state laboratory provides testing, and state personnel coordinate follow-up for women in state-supported prenatal clinics. Blood tests are free at the state health department laboratory for indigent patients receiving care at state clinics, but payment for follow-up services is the responsibility of either the woman or a third-party source. The program follows the ASHG guidelines. The public agencies assist private physicians and patients to obtain appropriate services from state-approved labs. Seventeen laboratories are state approved.

By regulation, Iowa restricts testing to one state-approved laboratory. All physicians can obtain the test for a fee of $27.50. The state also provides them with both expert consultation and assistance with interpretation and follow-up procedures. The state does not pay for follow-up or collect outcome data at this time.

In Washington, legislation was introduced in 1987 that would have required physicians to document the offering of AFP testing and that would have limited AFP testing to specially licensed laboratories. Regulations specifying the minimum number of tests, reporting requirements, and quality control would have been authorized. This bill was opposed by private laboratories and was amended to simply promote MSAFP testing and reporting. Nevertheless, it failed passage on the first attempt. The legislation was reintroduced and enacted in 1988. The state health agency now has regulations pending adoption. The regulations include laboratory standards which follow ASHG guidelines. The regulations also require that any health insurance offered in the state must include MSAFP as a benefit.

In 1988, after the first survey, Hawaii passed legislation giving the health department broad regulatory authority for all prenatal testing. Regulations to limit MSAFP testing to quality-controlled laboratories approved by the department are in the process of being adopted. The state health department has personnel assigned to collect data on outcomes and to assist with follow-up of positive tests.

Oregon was the only other state to adopt regulations after the first survey. Its existing laboratory-licensing regulations were modified to require the establishment of normal values for AFP testing, the adjustment of results for specific variables, and collection of detailed performance data.

The prenatal health committee of the Texas Medical Association sponsored a bill that would have given the Texas Department of Health the authority to pass regulations for MSAFP testing and to institute or require proficiency testing of laboratories providing the test. The bill was strongly opposed by the pathologists of the state, who viewed it as a "foot in the door," since currently the Texas Department of Health has no authority to regulate laboratories. The bill failed to pass its first committee hearing.

While no other state has enacted specific legislation or undertaken regulation of MSAFP testing, the public-health laboratories in four additional states (Nebraska, Kentucky, South Carolina, and Wisconsin) have offered limited MSAFP testing services—usually to public-health-clinic maternity patients who request testing. In the remaining 39 states, there was little state laboratory activity reported in either the 1987 or 1990 survey,

in terms of developing on-site analytical expertise, consultation, proficiency testing, or other AFP-related laboratory services.

In 1987 all but six state health agencies—i.e., those of Alaska, Delaware, Louisiana, North Dakota, Ohio, and Wyoming—were able to identify at least some of the commercial or university laboratories offering MSAFP testing, and most states reported that some specimens were sent to out-of-state laboratories. By 1990 only three states—Alaska, North Dakota, and South Dakota—could not identify a laboratory resource.

It is of interest that California was the only state reporting an ongoing effort to collect complete data on the number of MSAFP tests performed in the state. In 1987 only eight states felt confident enough to estimate the percentage of pregnant women screened, with these estimates ranging from 2% in Mississippi to 80% in Rhode Island. Most states reported having no knowledge of the extent, quality, or results of MSAFP testing at that time. In 1990 the estimated statewide coverage reported by each of 15 states ranged from 25% to 90%, but there are still no hard data collected, except in California, where 60% of women seen before 20 wk gestation are tested. A survey of MSAFP testing laboratories yielded an estimate that 1,000,000 (25.5%) of the 3,919,000 births had been screened in 1988 (Palomaki et al. 1990).

In spite of vigorous efforts to distribute the policy statement, 22 states in the 1987 survey stated that they were unaware of the existence of such standards. Twenty-three states reported that in-state MSAFP testing was meeting some but not all of the guidelines, and only five states indicated that all elements were being completely followed.

There was some improvement in 1990, since only 16 states reported ignorance of the ASHG policy statement. The turnover at state agencies resulted in some states being unaware in 1990 that had been aware in 1987 (see Appendix).

With respect to the proper organizational focus for addressing this kind of screening program, in 1987 eight states felt that it was primarily a state laboratory responsibility, while 27 states felt that it should be an MCH responsibility, and two felt that it should be a joint operation of both; three indicated that responsibility should be with crippled children's services (CCS), and 10 were uncertain as to where responsibility should be placed. By 1990 25 states said that responsibility should be with the MCH, nine said that it should be with the state laboratory, and seven said that it should be with the MCH and state laboratory combined; CCS, developmental disabilities, and genetics were each mentioned once, and six states were still undecided.

When asked to comment on the state's plans with respect to MSAFP in 1988, more than 30 states indicated that they had not given the matter any consideration. Several states commented that they were working on policy statements or that they were reviewing the problem. A few states (Michigan, Pennsylvania, Oregon, Virginia, and Florida) were considering active programs involving legislation, guidelines, or pilot projects. The legislation being considered at that time included reporting requirements, laboratory regulation, and fee-for-service testing.

In 1990 24 states still have no plans for MSAFP-related activity. When the states were asked to assign the issue a priority from 1 to 10, the results shown in table 1 were obtained.

Discussion

By its very nature, MSAFP testing is the kind of program that lends itself to a state or regional public-health approach (Fuhrmann 1985). The technical and regulatory issues involved have been reviewed elsewhere (Mizejewski 1987). To be effective, MSAFP testing must be available on a statewide basis, since there is no clearly definable high-risk group. A properly designed program also demands the skills of public education, assurance of quality laboratory work, accurate interpretation of test results, and public-health tracking to assure appropriate follow-up and testing of all positive results. Means and cutoffs must be based on a large population of pregnancies. For these reasons, experts have uniformly concurred with ASHG in recommending a centrally organized and regulated approach to protect against those abuses of the technology that have occurred in an uncontrolled environment (Haddow and Milunsky 1984; King's Fund Forum 1987; Royal College of Physicians 1989).

Table 1

Priority Assigned to MSAFP Testing by States

	Priority										
	0	1	2	3	4	5	6	7	8	9	10
No. of states[a]	1	5	2	4	3	5	3	6	9	2	2
Cumulative %	2	12	16	24	30	40	46	58	76	80	84

[a] For eight states, results are unknown.

The responses revealed the following four barriers to the optimal use of this new technology: lack of funding, lack of legislative authority, lack of state level expertise, and apparent lack of interest. These four barriers were present to differing degrees in individual states which did not have a formal program. MSAFP testing has been shown to be cost-effective in several studies, but the problem of redirecting some of the large amount of money spent on established treatment and rehabilitation programs to prevention remains a difficult one. While individual states may have difficulty funding a statewide screening effort, California's successful demonstration of a fee-supported statewide approach can be modified and duplicated in large states or in multistate regional programs.

Legislative authority for state agency involvement in regulating private laboratories is lacking in many states. Relevant laws need to be enacted or strengthened if MSAFP testing is to be used as the ASHG recommends. Some states have moved population-based testing, such as newborn screening, into state or academic multistate regional laboratories under contract, to achieve the economy of scale needed. This approach might be beneficially applied to MSAFP as well. There are several successful multistate newborn-screening programs that could be used as models for regional MSAFP programs. This regional approach to specialized tertiary genetic services has been endorsed by the U.S. Department of Health and Human Services MCH program in its funding and support of regional genetics networks.

State agencies will have to recruit experts in this area, as staff and/or consultants, to establish credibility in controlling such technology. In some instances the state health agencies have not yet embraced the area of genetic services as a public-health responsibility and have little or no interest beyond traditional basic public-health functions. If public health is to respond to tomorrow's challenges, this attitude will need to be changed.

Provision of well-developed policy statements by ASHG expert committees provides the starting point for practical development of high-quality comprehensive programs. Presumably, such statements are designed to encourage health agencies to play a more active role in the increasingly important area of genetic screening. To be effective, such statements need to be followed by more input and support, from local genetic experts to the state health agencies, and by more education of and coalition building with physicians, the public, and the state legislatures. Data and information need to be collected both on problems in uncontrolled testing and on benefits of organized programs by the genetic community. Meeting of local genetic experts with state health officials and obstetric providers should be held on a state-by-state basis to identify the barriers which prevent or delay implementation of such population-based programs. State agencies need to reassess their role in providing or regulating such genetic screening, since MSAFP is only the harbinger of other screening proposals for other genetic disorders.

Previous studies of the effect of policy statements on medical practice have demonstrated the necessity of adding incentives and removing disincentives by approprite actions of state health agencies, to encourage practitioners to adopt the recommended changes (Lomas et al. 1989). If the excellent work of the committees is to be more than an academic exercise, interaction with governmental agencies will be needed to promote rational genetic screening policies at the state level.

Appendix

State Health Agency Responses to ASHG Guidelines

Unaware of Guidelines, 1987:
- Alabama
- Colorado
- District of Columbia
- Georgia
- Illinois
- Indiana
- Kansas
- Louisiana
- Michigan
- Minnesota
- Mississippi
- Missouri
- Nevada
- New Mexico
- North Dakota
- Ohio
- Rhode Island
- South Dakota
- Texas
- Virginia
- West Virginia
- Wyoming

Unaware of Guidelines, 1990:
- Alabama
- Florida
- Kansas
- Louisiana
- Mississippi
- Missouri
- New Hampshire
- North Dakota
- Ohio
- Rhode Island
- South Carolina
- South Dakota
- Texas
- Utah
- Vermont
- West Virginia

References

American Society of Human Genetics, The (1987) American Society of Human Genetics policy statement for maternal serum alpha-fetoprotein screening programs and quality control for laboratories performing maternal serum and amniotic fluid alpha-fetoprotein assays. Am J Hum Genet 40:75–82

Garver KL (1989) Update on MSAFP policy statement from

The American Society of Human Genetics. Am J Hum Genet 45:332–334

Haddow JE, Milunsky AL (1984) Deregulation of screening for alpha fetoprotein in pregnancy. N Engl J Med 310:1669–1670

King's Fund Forum (1987) Consensus statement: screening for fetal and genetic abnormality. Br Med J 295:1551–1553

Lomas J, Anderson GM, Dominick-Pierre K, Vayda E, Enkin MW, Hannah WJ (1989) Do practice guidelines guide practice? the effect of a consensus statement on the practice of physicians. N Engl J Med 321:1306–1311

Main DM, Mennuti TM (1986) Neural tube defects: issues in prenatal diagnosis and counseling. Am J Obstet Gynecol 67:1

Mizejewski GJ (1987) Alpha fetoprotein testing: regulatory and technical considerations. Lab Management 25:19–28

Palomaki G, Knight GJ, Holman MS, Haddow JE (1990) Maternal serum alpha fetoprotein screening for fetal Down syndrome in the United States: results of a survey. Am J Obstet Gynecol 162:317–321

Royal College of Physicians (1989) Summary report: prenatal diagnosis and genetic screening. J R Coll Physicians Lond 23:215–219

Fuhrmann W (1985) Maternal serum alpha fetoprotein screening for neural tube defects: results of a consensus meeting. Prenat Diagn 5:77–83

Tosi LL, Detsky AS, Roye DP, Morden ML (1987) When does mass screening for open neural tube defects in low-risk pregnancies result in cost savings? Can Med Assoc J 136:255

Wald NJ, Cuckle HS (1987) Recent advances in screening for neural tube defects and Down syndrome. Baillieres Clin Obstet Gynaecol 1:3:649–676