Newborn screening has traditionally referred to biochemical testing for inherited disorders, generally metabolic in origin, that are usually correctable by dietary or drug interventions. As new tests have been developed, state public health newborn screening systems have slowly evolved without the benefit of national policies. Thus, newborn screening program changes, when viewed nationally, have been uncoordinated. The net result has been unequally applied mandated screening and, consequently, unequal availability of related public health disease prevention services. Technological advances in laboratory testing over the past 10 years have resulted in limited program changes in some state newborn screening systems, and even greater program disparities. A recent Newborn Screening Task Force identified numerous issues of concern and proposed elements for a plan of action involving public health programs, healthcare providers, and consumers. This minireview details past policy history in newborn screening and identifies some of the current issues confronting programs as they seek to move ahead with the technologies and medical treatments for the twenty-first century.

Key Words: newborn screening; policy; tandem mass spectrometry; DNA; public health.
state government involvement to ensure quality and availability of screening services for all newborns, to conservative state government spending policies, and to the lack of national policies, absent a national law, related to the various aspects of newborn screening and newborn screening systems.

Without a national newborn screening policy or models for guidance, newborn screening programs have expanded sporadically over the years, sometimes as a result of scientific findings (e.g., new testing technologies, automated punching), sometimes due to financial incentives (e.g., federal support of sickle cell testing), and sometimes due to consumer advocacy and accompanying political pressures. Concerned consumer advocates directly influenced early screening efforts and consumers have continued to influence some state programs by occasionally encouraging addition of disorders to the screening program. Currently, as a result of scientific and technological advances, and because of increased consumer interest, state governments are experiencing increased attention to expanding state mandated newborn screening for certain rare metabolic disorders detectable through tandem mass spectrometry testing.

Other programs and technologies are also impacting newborn screening. Technological advances in newborn hearing screening procedures are now bringing to fruition the long-range goal of screening all newborns for hearing impairment discussed since the early 1960s (11). These programs are generally developing apart from existing newborn screening programs often without the benefit of their experiences (particularly in follow-up and service delivery). Output from the human genome project has the potential for impacting newborn screening through multimutational "chips" that could be used in genotypic analysis of newborn screening disorders, at least for confirmatory testing (12,13). New tests are also allowing detection in newborns of genetic predispositions to conditions not manifested until later in life (14,15). Each new testing possibility raises again the legal and ethical questions of who should know this genetic information, what they should know, and where the knowledge should reside. For newborn screening programs, there are also the continuing concerns of patient's rights—particularly the right to information about testing and the right to have personal information private and protected from misuse.

BACKGROUND

In order to understand the public policy issues involved in mandating population newborn screening, it is important to consider the history underlying current policy (16,17). Initially there was not wide acceptance of newborn screening for PKU within the medical community because of the relative rarity of the disorder. Guthrie, who had a mentally handicapped son and a niece with PKU, was active in community support groups for the mentally handicapped and took his screening ideas to these groups for the political support needed to move screening ideas into government-run public health institutions. As parents of mentally handicapped children learned how newborn screening could detect some disorders causing mental retardation, and that early detection and treatment could limit the effects of mental retardation, and thus reduce both the psychological and economic burdens to families, they exerted the necessary political pressures that led health departments to begin universal newborn screening programs. In 1965, the American Academy of Pediatrics Committee on the Fetus and Newborn finally recommended a newborn screening blood test for PKU for all newborns “no sooner than 24 hours after onset of milk feeding and prior to discharge" (18).

Federal government funding through the Children's Health Bureau [now the Maternal and Child Health Bureau (MCHB) of the Health Resources and Services Administration (HRSA)] supported the early studies by Guthrie, as well as the health promotion that followed. As state programs were established, they were usually funded from tax monies and the only charges to parents, if any, were those imposed by the collecting hospital for administering the program. However, some hospital and private laboratories saw this testing as potentially profitable and also offered newborn screening testing for a fee (often included as part of the maternity charge). Unfortunately, because of the low incidence of the disorders in question and the quality control issues involved in rare disorder testing, cases occasionally went undetected. Eventually liability concerns caused most private testing laboratories to reconsider their testing strategies and defer testing to state public health laboratories. Screening laboratories developed into screening systems that included follow-up and education, not always as part of the laboratory infrastructure. In sparsely populated states, it was more efficient to regionalize the labo-
ratory part of the screening system, and thus three major regional laboratories initially arose in Massachusetts, Colorado, and Oregon. Over the years, some other states formed partnerships, including North Dakota and Iowa, Mississippi and Tennessee, and Delaware and Maryland. As time went by, some of these arrangements shifted, but regional laboratories still remain in Massachusetts (Massachusetts, Maine, New Hampshire, Vermont, and Rhode Island) and Oregon (Oregon, Alaska, Hawaii, Idaho, and Nevada), with partnerships remaining with North Dakota/Iowa, Colorado/Wyoming, and Tennessee/Mississippi. One large private laboratory currently contracts to provide testing services in the District of Columbia and Pennsylvania.

With time, state legislatures became more fiscally conservative and most required state-funded programs and services to become financially self-supporting. Thus, the funding mechanism for newborn screening in most states evolved into fee-for-service. Today, newborn screening programs remain state-based with wide variations in their funding, formulation, and administration. Despite the wide ranging program variability, state public health newborn screening systems have been considered successful in having created efficient, productive disease detection and service delivery systems for thousands of newborns identified annually with heritable disorders.

Federal support for newborn screening has continued through the years in various ways. Most visible has been the funding of various working groups, educational symposia, special projects of regional and national significance by HRSA, various consensus conferences by the National Institutes of Health (NIH), and national laboratory quality assurance and education efforts by the Centers for Disease Control (CDC). Cooperative efforts between federal agencies, in support of newborn screening, have also positively impacted newborn screening and helped shape limited national policies. In 1987, for example, NIH convened a consensus conference on newborn screening for sickle cell disease and other hemoglobinopathies to address the questions essential to developing national newborn screening guidelines in this area. The resultant consensus statement (19) concluded that newborn screening for sickle cell disease should be universal (as opposed to targeted) utilizing the concept of centralized laboratories, and that screening programs must include comprehensive care in their service considerations. Answering the funding dilemma facing many states considering implementation of these guidelines, HRSA provided $1.3 million in newborn screening program grants in 1988, $3.88 million in 1989, and $6.86 million in 1990 to assist in implementing newborn screening for sickle cell disease in states needing funding assistance (20) in order to implement the guidance resulting from the NIH conference.

More recently, states have been confronted with questions about adding other screening tests for disorders for which cures do not currently exist or for which treatments may not yet be established. Federal support has again been forthcoming to aid in the scientific debates that may eventually lead to national policies. One example is a 1997 CDC-sponsored workshop (21) on newborn screening for cystic fibrosis (CF) “to discuss the benefits and risks associated with screening newborns for CF and to develop public health policy concerning such screening.” In considering a national newborn screening policy regarding CF, participants agreed that, “before recommending universal CF screening for newborns as a routine public health intervention, policymakers will need more compelling data about its effectiveness.” Included in the report were suggestions for further studies and plans for reviewing data and arguments within 2 years, using a national consensus panel.

In 2000, the National Newborn Screening and Genetics Resource Center (NNSGRC), with HRSA funding and CDC program development assistance, sponsored a working group meeting of experts to expertly review the issues of implementing tandem mass spectrometry (MS/MS) testing as a means of screening newborns for rare metabolic diseases (22). This working group, while not recommending a national policy on the use of MS/MS, nonetheless began the policy-making process by making “proposals for planning, operating and evaluating MS/MS for analyzing dried blood spots routinely collected from newborns.” This group did not address addition of specific disorders to newborn screening programs, but rather addressed issues and impediments to technology implementation. Because of the intense consumer interest in expanding newborn screening programs, and the potential for exchanges of inaccurate information among the consumer community, the participants agreed that, “the public should receive accurate information regarding expanded and comprehensive newborn screening and the evolving knowledge regarding its strengths and weaknesses.” Ongoing screening programs were encouraged to continue acquiring and sharing scien-
tific data, and emphasis was placed on including other appropriate, and sometimes overlooked, disorders (besides those of metabolic origin) in comprehensive screening programs (e.g., congenital adrenal hyperplasia, cystic fibrosis, sickle cell disease, and biotinidase deficiency).

CURRENT SCREENING ENVIRONMENT

Newborn screening is a SYSTEM composed of six parts, five of which have been discussed in detail elsewhere: screening, follow-up, diagnosis, management, and evaluation. The sixth, education, permeates the system and provides the mechanism for enhancing all other system components. Newborn screening is a system that must function within geographic, economic, and political constraints, and which must smoothly integrate sample collection, laboratory analysis, follow-up, diagnosis, and treatment. There are approximately 4 million births annually in the United States and essentially all undergo some form of newborn screening, although there is extreme variability in the way in which screening occurs.

All states and the District of Columbia have legislation that creates a newborn screening program. In most, the program is placed in, or under the supervision of, the state health department. Decisions about addition or deletion of newborn screening tests and other critical issues, including funding, are usually left to the state health officer or his/her designee. Many newborn screening programs have an advisory committee (or in some cases subspecialty committees such as metabolic, hematology, endocrinology) that provides input into program operations. In these programs, changes are usually evaluated by the committee in consultation with newborn screening program personnel, and recommendations are made to the state health officer or board of health. There is currently no national model for enacting program changes and most programs do not have formal procedures for adding or subtracting tests, and thus, changes in screening testing panels may occur in any number of ways. In some cases they result from published scientific studies or other science-based research. In other cases they may arise because of political or financial considerations, sometimes unrelated to the impact of the disorder on improving the public's health.

The number of mandated screening disorders included in each state screening program varies from 3 to 21 (as of this writing, Wisconsin requires 21 disorders), and several states are contemplating adding more (e.g., Minnesota, New Jersey, New York). Some states (e.g., Massachusetts, Maine, South Dakota) offer optional biochemical testing for more than 30 disorders, and various supplemental newborn screening tests are available from several commercial or not-for-profit laboratories. All but 6 states charge a fee for newborn screening, although not all apply the fee directly to newborn screening program expenses, and not all fees are calculated in the same manner or cover the same services. State newborn screening patient charges vary from $0 to $60, with hospital administrative fees often an additional expense on the patient's bill. The comparability, or lack thereof, of state newborn screening fees cannot be directly linked to the number of tests offered. Essentially all states maintain a newborn screening database of patients tested biochemically and track presumptive positive patients at least until the point of diagnosis and treatment. Many are developing newborn hearing screening programs and are considering data integration to avoid duplication of data collection efforts, to improve statistical data collection, and to track diagnostic and treatment service delivery better. Almost all state newborn screening collection cards utilize some form of serial number for inventory control with the potential for utilization as a linking number between public health databases. Collection cards containing residual blood, after testing is completed, are usually stored for specified periods of time varying from 2 weeks to indefinitely. Storage conditions are variable including many laboratories who store samples in filing boxes at room temperature and some who use refrigerated storage with desiccant in order to minimize adverse effects of humidity. There is not yet a national consensus regarding appropriate length of storage, storage conditions, and appropriate uses beyond newborn screening, nor is there consensus concerning consent for storage and usage, although the issues requiring consideration have been enumerated.

POLICY FORMULATION

With the evolution of newborn screening in the early 1960s came the need for discussions concerning not only technical details of screening procedures and disease treatment protocols, but also screening policy. Three policy documents have had the greatest impact on U.S. newborn screening programs through the years. The earliest of these was
in 1967, and resulted from a World Health Organization (WHO) Scientific Group on Screening for Inborn Errors of Metabolism that was convened in Geneva, Switzerland, with the purpose of considering, “whether and how newborn screening programs could improve the health of mankind.” The published proceedings of this meeting (28) reference five companion WHO reports from 1964 to 1968, as pertinent to various aspects of the topic. The WHO group divided screening conditions into three groups: “(a) conditions for which there is a well-defined screening test and a fairly uniform policy of management once the disease has been identified; (b) conditions in which the abnormal gene can regularly be identified but in which the condition only becomes symptomatic in a specific environment; and (c) a miscellaneous category which includes conditions for which more information is needed before they will fit easily into a routine screening program.” Final recommendations of the WHO Scientific Group are given in Table 1.

One of the companion reports identified by the WHO Scientific Group, the report by Wilson and Jungner (29), is perhaps the most referenced source of screening criteria. Their “Principles of Early Disease Detection” (see Table 2) were an attempt to chart the path for public health agencies to take in “bringing to treatment those with previously undetected disease” while “avoiding harm to those persons not in need of treatment.” Wilson and Jungner noted that early screeners for PKU did not anticipate problems with acceptance of newborn screening and so, did a poor job of documenting their successes. Because of the need for scientific documentation and science-based information, when evaluating new screening programs, they enumerated detailed recommendations (paraphrased in Table 3) regarding newborn screening, keeping in mind the idea that rigidity was useful for guidance but should not stifle initiative.

In 1974 Frankenburg (30) revisited these screening principles in discussing how physicians might best decide on diseases to be included in private practice screening programs, given the rapid expansion and availability of new screening tests (Frankenburg’s criteria are summarized in Table 4). In defining these criteria he noted that, “if the decision to screen is based only on the availability of a test or the apparent importance of a test, great harm can be done, both in the waste of valuable medical resources and in direct harm to the persons screened.” Further, he concluded that, “The availability of a

### Table 1
**Recommendations—WHO Scientific Group, 1968 (28)**

1. Appropriate techniques and methods should be developed for screening general populations as well as high-risk groups for certain inborn errors of metabolism.
2. Automatic procedures should be developed for the analysis of samples and handling of data.
3. The long-term storage of biological specimens should be studied.
4. Large-scale pilot studies should be made to evaluate and compare screening methods.
5. Selected populations should be investigated to obtain data on the frequency of these diseases and traits.
6. Multidisciplinary groups should be set up to study the short-term and long-term social and biological consequences of screening programs.
7. In each proposed screening program, a careful estimate should be made of the cost of the program and of the personnel, facilities, and equipment required.
8. Central laboratories specialized in screening procedures should be created on a regional basis and those that already exist should be assisted.
9. Specialized regional centers should be set up for the study and the management of patients or, if they exist, expanded.
10. Collaborative studies should be undertaken to evaluate the investigation and management of patients.
11. International cooperation should be enlisted for the exchange and training of personnel, the exchange of information and materials, and the comparison of methods and results.

### Table 2
**Principles of Early Disease Detection—Wilson and Jungner, 1968 (29)**

1. The condition sought should be an important health problem.
2. There should be an accepted treatment for patients with recognized disease.
3. Facilities for diagnosis and treatment should be available.
4. There should be a recognizable latent or early symptomatic state.
5. There should be a suitable test or examination.
6. The test or examination should be acceptable to the population.
7. The natural history of the condition, including development from latent to declared disease, should be adequately understood.
8. There should be an agreed policy on whom to treat as patients.
9. The cost of case finding (including diagnosis and treatment of patients diagnosed) should be economically balanced in relation to possible expenditure on medical care as a whole.
10. Case-finding should be a continuing process and not a “once and for all” project.
All newborns should be screened for PKU, using blood, as late as possible before hospital discharge, and other disorders for which immediate intervention is imperative should be combined in the screening process.

When tested too early for PKU (<5 days after birth), the baby must be tested again after 4 weeks, and this testing might be combined with other disorders for which detection from a late screen might be preferable or for which there is no evidence supporting the need for immediate intervention.

Greater quality control of laboratory testing is essential and, therefore, testing should be performed in large state or regional laboratories and not in small state or private settings. The number of live births tested should be the prime determinant of the size of the region while taking into account geography and state borders.

A single laboratory (probably at the CDC) should have responsibility for maintaining proficiency of the testing laboratories.

There should be greater standardization and efficiency in reporting results and in maintaining results for statistical purposes.

Patients should be referred to specialty centers for diagnosis and the testing laboratory should exhibit proficiency in the type of testing used. Family physicians should be kept informed of the outcome of referred patients.

If screening of all newborns is mandated, then there exists an obligation to ensure optimal therapy. Adequate financing must be available and a societal responsibility exists when families are unable to pay.

Treatment should be continually improved, with government support if private industry is unable to adequately pursue.

Since there is a high risk of retardation to the offspring of mothers with PKU, screening for PKU should be a part of early antenatal care (if screening status is unknown). Until the efficacy of a low protein diet during pregnancy is proven, termination of pregnancy should remain an option.

Legislative decisions about newborn screening should be science based and the benefits realized should outweigh the costs. In order to avoid fragmented, uneducated, and hurried decision-making, there should be advisory bodies with ongoing responsibility and competence in the field of genetic screening, including both medical and non-medical expertise.

The components of a successful program should include: prescreening professional and public education, properly timed tests, centralized laboratories, careful quality control, rapid follow-up, state-sponsored medical and nursing consultation for families, and free treatment.

Parents should be aware of their right to refuse testing at an early enough time to exercise that right.

**TABLE 3**

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</table>

Suitable screening test does not justify screening for a disease unless the disease is important, relatively prevalent, and amenable to early treatment. Screening for a disease which has the necessary characteristics cannot be justified unless there is an acceptable, reliable, and valid test which can be carried out at reasonable cost. Screening which is carried out without knowledge and consideration of these criteria is likely to be wasteful of scarce medical resources and may actually do more harm than good.

The second major policy report was published in 1975 by the National Academy of Sciences, having been commissioned by the Division of Medical Sciences of the National Research Council at the request of the Social Issues Committee of the American Society of Human Genetics, issued its report on genetic screening (31). This report, in response to rapid advances in genetics and its possible future impact, was intended to "review current screening practices . . . identify the problems and difficulties and give some procedural guidance, in order to minimize the shortcomings and maximize the effectiveness of future genetic screening programs." This comprehensive 8-part report produced 21 recommendations that were divided into the 5 categories as summarized in Table 5. This report and its rec-

**TABLE 4**

<table>
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<th>Selection Criteria for Screening—Frankenburg, 1974 (30)</th>
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<tbody>
<tr>
<td>1. Importance of the problem—The disease or condition should be serious or potentially serious, and to be cost effective, should have a positive impact on morbidity and mortality.</td>
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<tr>
<td>2. Prevalence—The condition should be relatively common or prevalent (in order to lower the cost of case detection).</td>
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<tr>
<td>3. Accepted criteria for diagnosis—It must be possible to diagnostically differentiate diseased from non-diseased individuals. Because screening invariably presents problematic cases intermediate between disease and non-disease and thus, it is essential to define (as precisely as possible) what is to be identified by screening.</td>
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<td>4. Treatable or controllable—It should be possible to reverse, slow the disease process or ameliorate the disease. In certain non-treatable hereditary conditions, screening offers the opportunity for informed decisions as part of family planning.</td>
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<td>5. Advantage of earlier treatment—Treatment resulting from early screening detection should be more effective or efficient than if delayed until the usual time of diagnosis.</td>
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<td>6. Adequate time from screening to treatment—There must be sufficient time available between the time of the screening test and the optimal time for treatment.</td>
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<tr>
<td>7. Availability of diagnostic and treatment resources—There must be sufficient funds, facilities and personnel available to diagnosis and treatment of all individuals with positive screening test findings.</td>
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<tr>
<td>8. Program cost—The cost of asymptomatic screening, diagnosis, and treatment should be outweighed by the savings in human misery and fiscal expenditures if the disorder is not detected until symptoms occur, and the cost must be justified relative to other ways medical resources might be deployed.</td>
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TABLE 5
National Academy of Sciences Recommendations—1975 (31)

1. General—Screening is appropriate when carried out in controlled conditions when certain criteria are met: there is evidence of substantial benefit and acceptance; feasibility has been investigated and benefits outweigh costs; appropriate public education can be accomplished; test methods are satisfactory; laboratory facilities are available; and resources exist to deal with counseling, follow-up, and other testing consequences. PKU screening should be continued and the experiences gained (positive and negative) should be considered in the design of future programs.

2. Organizational—Screening responsibility should be in an agency representing both the public and health professions because of the public nature of screening and the potential for invasion of privacy. Public representation is necessary in determining public good and designing population screening programs. There should be clear aims of screening programs, consultation with medical societies, and standardization of protocols. Programs should include quality control and should be regional based on population numbers rather than political subdivisions. Genetic screening should be one part of preventive health measures dictated by general principles governing genetic screening rather than by pressures originating in the special qualities of particular diseases.

3. Educational—Studies of human biology, including genetics and probability, should be started in primary school and effectiveness of public education should be improved through use of mass media and other techniques of proven value. Continuing education for physicians should emphasize human genetics and practical application of population genetics, and medical schools should include genetics education in courses in epidemiology, preventive medicine, medicine pediatrics and obstetrics. Educational institutions should receive support for programs to set standards and train personnel for information and counseling for screening programs.

4. Legal—Screening should not be mandatory and privacy should be protected. Screening authorities should consult regularly with lawyers and ethicists to ensure that there is no legal or social damage occurring as a result of screening. When mandatory screening is considered, it is recommended that consideration be given to a law creating a Board on Hereditary Disorders such as that proposed by the Council of Pediatrics (AAP) in 1999. The resulting report (32) was the third major policy report impacting U.S. newborn screening. This task force responded to two primary tasks: (1) to review the issues facing state newborn screening systems, and (2) to make recommendations. The task force consisted of multidisciplinary groups comprising 5 teams working to develop the final report. The five workgroups included: (1) newborn screening and its role in public health; (2) medical home and systems of care; (3) economics of screening; (4) ethical, legal, and social issues; and (5) research, surveillance, and assessment issues.

In defining the issues, the Task Force first described its “basic assumptions” about newborn screening:

1. Infants should benefit from and be protected by newborn screening systems.
2. Using previously defined (WHO) criteria for inclusion of a screening test, not all conditions are good candidates for newborn screening.
3. Newborn screening is a system and every newborn should receive appropriate and timely services.
4. Newborn screening is an essential public health prevention activity requiring service integration for affected newborns.
5. State public health agencies have responsibility for assessment, assurance, and policy development.
6. The newborn screening system must be clinically, socially, and ethically acceptable to the public and health professionals.
7. Every newborn should have a medical home.
8. All newborns should have access to screening according to nationally accepted criteria regardless of their location.
9. Parents have a right to information about newborn screening, the right to refuse testing, and the right to privacy protection.
10. Increased newborn screening program coordination and uniformity will benefit families, healthcare professionals, and public health agencies.
11. Parents and consumers must be involved in policymaking and program implementation.

The Task Force made recommendations in 4 ma-
jor categories: (1) public health infrastructure; (2) public and professional involvement; (3) surveillance and research; and (4) financing. Foremost among their concerns was the issue of unequal availability of state screening services depending on the state in which the birth occurs, and the possibility of a national policy defining the parameters for state-mandated testing programs. Because state newborn screening services are inextricably linked to funding and state economies are varied, program finances were also a major concern. In summary, an agenda for action was proposed for a partnership among the public health system(s), health professionals, and consumers to continue a process that:

1. Defines responsibilities—federal and state.
2. Models regulations for newborn screening systems.
3. Defines minimum standards for newborn screening systems.
5. Models systems of care from infancy to adulthood.
6. Designs strategies to inform and involve families and the general public.
7. Funds demonstration projects to evaluate technology, quality assurance, and health outcomes.

Publication of the Task Force Report elicited a press release response from the March of Dimes Birth Defects Foundation (MOD) in which all state newborn screening programs were encouraged to mandate screening for 8 disorders: PKU, congenital hypothyroidism, congenital adrenal hyperplasia, galactosemia, sickle cell diseases, biotinidase deficiency, maple syrup urine disease, and homocystinuria. This was followed later by a published commentary (33) in which testing cost was identified as an unnecessary element in newborn screening policy decisions. MOD also took the position that newborn screening (even for rare diseases) should be conducted on every newborn, “as long as its early discovery makes a difference to the child.”

Continuing technological advances and outfall from the successes of the Human Genome Project have heightened awareness of two important questions: (1) how should states best provide state-of-the-art screening; and (2) how should newborn screening programs best cope with the ethical, legal, and social implications of expanded testing that might mean detection of disorders for which no, or at best limited, treatment exists. These newborn screening policy issues are compounded by the availability of expanded testing in the private sector and its marketing to prospective parents eager to have the best possible health outcome (and therefore all available newborn screening procedures) for their newborn. Availability of private testing and the need for access to public health services routinely provided as follow-up to disorders detected through state newborn screening programs presents new challenges to state public health systems. Despite the availability of screening tests from private laboratories, state legislatures are being asked to mandate expanded newborn testing, often without considering the impact of expanded testing on other system components, including follow-up of presumptive positive laboratory results and the expert laboratory and medical evaluations and personnel necessary for proper diagnosis and treatment. Without national policies on newborn screening, each state continues to develop its own approach.

Concerted efforts by consumer advocacy groups, at least partially resulting from commercial marketing of private supplemental newborn screening testing (primarily using MS/MS technology and offered as an option in some hospitals, particularly in Pennsylvania), led to a media awareness campaign during 2000. The result was, and continues to be, a proliferation of newspaper and magazine articles related to newborn screening. Responding to these and other consumer requests, several state legislatures have now passed more comprehensive newborn screening legislation. In Mississippi, for example, it is now required that parents be informed prenatally and postnatally of the availability of screening tests for disorders not included in the state mandated newborn screening program (34). A
joint resolution by the 92nd Assembly of Illinois (35) encouraged the Illinois Director of Public Health and the Director of the Illinois Newborn Screening Program to adopt an information policy about other available newborn screening tests similar in intent to the Mississippi law. In Missouri, newborn screening was legislatively redefined recently to include a large number of metabolic and other disorders (36). In New Jersey, the governor convened the State Newborn Screening Advisory Committee to investigate and make recommendations on which he immediately acted to expand the New Jersey newborn screening testing battery from four to fourteen. Further, he reconvened the group to consider additional testing almost immediately after their initial report was completed. Many other states are considering expanding their screening programs to include as many as 30 disorders, some using federal funding to develop models for use in other states.

**DISCUSSION**

In newborn screening systems, there are critical junctures within and between system components where responsibilities and coordinated activities must be seamless and nonduplicative. Defined beginning and ending points for system components are critical in ensuring that all patients with presumptively identified disorders through newborn screening receive full advantages of the screening and public health system. Partnerships between public and privately funded components must be directed toward achieving the goal of fast and effective diagnosis and treatment for all identified patients. Newborns are already receiving supplemental screening in many states through private screening laboratories. These screening activities are often independent of state public health systems, including follow-up and data coordination, and they are usually unavailable to those who cannot afford them. Unless there is a cooperative mechanism for appropriate follow-up of patient results resulting from such testing to the point of timely diagnosis and treatment, a newborn screening system cannot function properly. Additionally, universal availability of newborn screening requires a funding mechanism for those unable to pay. The benefits of early screening may be compromised when there is competition between private and public newborn screening programs and a comprehensive, functional and efficient newborn screening system becomes fragmented.

Because screening almost always detects patients with variable clinical responses to the disorder screened, there will always be the need for interfaces with trained diagnosticians in order to appropriately determine which presumptive patients are actually affected, which of these may need treatment, and to what extent. These types of follow-up have traditionally been available through government-sponsored newborn screening programs, but have been limited to support of the government-mandated testing disorders. If the public health follow-up support system absorbs the follow-up from private testing, can it maintain the quality of follow-up for mandated testing? If not, how can a public/private partnership be created to address this problem? Similarly, if data are needed within the screening system in order to evaluate its operation and effectiveness, and these data exist in private screening systems and in public screening systems, how can they best be combined? How can patient privacy be assured in a data-sharing arrangement between the public and the private sectors? How can data integrity be assured? Data for these purposes must flow seamlessly and efficiently through the newborn screening system while maintaining the full integrity of the system. Follow-up must feed into diagnosis and treatment in the same way. Care must be taken to avoid destructive competitions between public and private system components through education, evaluation, and carefully defined responsibilities. Ultimately, public health departments must ensure that newborn screening systems function to the benefit of the public's health.

Payment for treatment services, such as dietary supplements, has been a continuing issue since PKU screening began. The question of payment for treatment services will only grow as screening expands. While newborn screening occurs shortly after birth, it detects conditions that must be dealt with over a lifetime. While some disorders require prophylactic medications, others require surgery or lifelong dietary intervention, all at a cost. Dietary intervention and supplementation, which are usually relatively expensive, are particularly troublesome because insurers fail to correlate medically necessary foods with medical treatment. Future policies should account for financial issues resulting from transitioning from childhood to adulthood. Insurers must receive better education about the long-term benefits of newborn screening before they will understand the need for treatment coverage beyond childhood.

Lessons learned from newborn screening imple-
NEWWBORN SCREENING POLICY DILEMMAS

In establishing newborn screening policies for the good of the public's health, there are many dilemmas and questions that require renewed attention within the advancing genetic testing environment of the twenty-first century. For example, are any of the WHO criteria of the 1960s still appropriate? If so, which ones? If not, why not and how should they be changed? Has new technology redefined the criteria for screening such that screening for families of disorders rather than individual diseases should be the criteria used for looking at incidence, cost, and system effectiveness? If this is the case, as some have suggested, what is the optimal way to approach disorders for which cures are not available? Is there a role for newborn screening in defining causes of fatal genetic disorders relative to family planning (37)?

Should newborn screening samples be used for other purposes such as forensics, and if so what are the parameters (38,39)? How is patient privacy best protected? How are decisions about adding and deleting screening tests best made (24)? Can disorders be prioritized for addition to screening programs and if so, by whom and how? What are the parameters for successful shared decision-making and how can this be successfully integrated into newborn screening programs? What are the components of the successful newborn screening system and how are they financed? How is treatment best financed and how is treatment financing transitioned from childhood to adulthood?

The AAP Task Force Report laid the groundwork for needed actions. Some of these are already underway with federal financial support. HRSA has established contracts for policy development in the area of retention and use of residual newborn screening blood spots. Similarly, a contract has been established for reviewing issues of equal access to screening tests. The CDC has been provided with funds to incorporate newborn screening activities beyond its current newborn screening quality assurance efforts, and hopefully these activities will complement the policy development efforts of HRSA. Newborn screening is a system of early identification of genetic and other health problems that impact families and its benefits must continually be reevaluated and strengthened.

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