

Serving the Family From Birth to the Medical Home

Newborn Screening: A Blueprint for the Future

A Call for a National Agenda on State Newborn Screening Programs

ABBREVIATIONS. PKU, phenylketonuria; AAP, American Academy of Pediatrics; MCHB, Maternal and Child Health Bureau; ARC, Association for Retarded Citizens; MCH, maternal and child health (programs); CDC, Centers for Disease Control and Prevention; HRSA, Health Resources and Services Administration; CSHCN, Children With Special Health Care Needs; CORN, Council of Regional Networks for Genetic Services; NAS, National Academy of Sciences; IOM, Institute of Medicine; HHS, US Department of Health and Human Services; NIH, National Institutes of Health; AHRQ, Agency for Healthcare Research and Quality; APHL, Association of Public Health Laboratories; SACGT, Secretary's Advisory Committee on Genetic Testing; CLIA, Clinical Laboratory Improvement Amendments; HCFA, Health Care Financing Administration; NSQAP, Newborn Screening Quality Assurance Program; WIC, Supplemental Nutrition Program for Women, Infants, and Children; HEDIS, Health Plan Employer Data and Information Set; NBAC, National Bioethics Advisory Commission; IRB, institutional review board; OTA, US Congress Office of Technology Assessment; SSI, Supplemental Security Income; SCHIP, State Children's Health Insurance Program; HIPAA, Health Insurance Portability and Accountability Act; ERISA, Employee Retirement and Income Security Act; EPSDT, Early and Periodic Screening, Diagnosis, and Treatment (program).

I. BACKGROUND

Newborn screening in the United States is a public health program aimed at the early identification of conditions for which early and timely interventions can lead to the elimination or reduction of associated mortality, morbidity, and disabilities. This screening takes place within the context of a newborn screening system, and involves the following components: screening, short-term follow-up, diagnosis, treatment/management, and evaluation. Inherent to each of these components is an education process.

The screening programs like these for the 4 million infants born each year in the United States have been heralded as successful and cost-effective.¹⁻⁵ The newborn screening program's efficiency and effectiveness depends on the smooth integration of sample collection, laboratory testing, follow-up, diagno-

sis, timely treatment, and tracking of outcomes.⁶⁻¹¹ The foundation and justification of newborn screening systems rest on the principles that testing procedures are readily available; technically feasible; economically sound; and clearly beneficial to affected newborns, their families, and to society.^{10,12-14} The universal acceptance of newborn screening for specified conditions over the past 3 decades attests to the undeniable benefits that flow from early testing and prompt, appropriate therapy. However, although newborn screening systems have succeeded in preventing morbidity and mortality, controversies, challenges, and opportunities continue.

The History of Newborn Screening

Newborn screening programs began in the early 1960s with the original work of Dr Robert Guthrie, who developed a screening test for phenylketonuria (PKU) and a system for collection and transportation of blood samples on filter paper.^{15,16} By 1962, Massachusetts launched a voluntary newborn PKU screening program that demonstrated the feasibility of mass genetic screening.¹⁷

Initially, newborn screening for PKU was not a health department role or a legislated activity. Health professionals were slow to adopt the practice of screening for PKU, and the responsibility for screening was not defined (eg, should it be the responsibility of the hospital in which the infant was born, the mother's obstetrician, or the infant's pediatrician or primary care health professional). The American Academy of Pediatrics (AAP), acting as the professional association that develops policy for the care of children, raised concerns about the sensitivity and specificity of PKU screening tests, as well as the efficacy of early intervention for PKU.^{16,17} Out of these concerns, the need for further research about this testing was recognized, and the federal Children's Bureau (now the federal Maternal and Child Health Bureau [MCHB]) funded a collaborative study to address questions and concerns about the effectiveness of the PKU screening test.^{16,17}

At the same time, advocates for children remained concerned that children with undetected PKU were at high risk for mental retardation.^{16,17} The National Association for Retarded Citizens (now the ARC)

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proposed model legislation for creation of public programs to address low detection rates, and also conducted an extensive grass-roots lobbying effort to support passage of mandatory PKU screening legislation.¹⁶ Many state health departments supported the adoption of such legislation. The Kennedy Administration, with the guidance of the Presidential Advisory Commission on Mental Retardation, was also supportive. The Commission hired the Advertising Council, which mounted a public campaign for mandatory PKU screening. Other advocacy groups, such as the March of Dimes Birth Defects Foundation, mobilized volunteers to lobby for passage of legislation at the state level. As a result of this multidimensional advocacy campaign, most states passed laws in the early 1960s that mandated newborn screening for PKU.^{17–19} Forty-three states had formal statutes by 1973. State health departments, particularly their maternal and child health (MCH) programs (funded by Title V of the Social Security Act of 1935), assumed the central role in implementation of these new laws.^{16,17}

As a response to this mandate, some states set up screening laboratories or added phenylalanine analysis to their state laboratory's repertoire of tests. In other states, private laboratories played a major role. Quality control was difficult because of the number of and the variability among testing sites; and became even more difficult as states added other genetic tests to their newborn screening batteries. Early in the 1970s, the need to improve quality assurance through systematic proficiency testing was recognized. In an early proficiency-testing study, the Centers for Disease Control and Prevention (CDC) found marked variability among health department laboratories. As a result, the Newborn Screening Quality Assurance Program was begun at the CDC, with additional funding from the Health Resources and Services Administration (HRSA).²⁰ (See further discussion in Section II, Public Health Infrastructure.)

In 1976, federal legislation to support screening for genetic diseases was adopted, and in fiscal years 1979 and 1980, 34 state genetic service programs received federal funding.^{21,22} This support was welcomed by the states, as the cost of screening tests and the health departments' coordination of screening activities had not been completely covered by many state budgets.

As a result of the laws mandating PKU testing, and the establishment of health department newborn screening units that occurred in the 1960s and 1970s:

- Every newborn had an opportunity to be screened for PKU when laws were properly implemented; consequently, most were screened.
- Financial barriers to screening and diagnosis were removed, but families often had to pay for the special formula, special foods, and other related treatments.
- State newborn screening programs evolved, with the goal of providing safe screening tests and appropriate follow-up to every newborn.¹⁷

During the 1980s, further systems development took place at the state and regional level. Newborn

screening systems were set up by public health agencies to ensure coordination between the hospitals from which most specimens were received, the public health laboratory, the infant's pediatrician or primary care health professional to whom positive results were reported, and pediatric subspecialists to whom infants were referred for diagnosis and treatment.^{7,12,23–26} Together these entities comprised the backbone of newborn screening systems. Some state newborn screening systems also played a role coordinating follow-up^{25,27}; depending on their public health structure, medical care structure, and available resources. In many states, the Title V Children With Special Health Care Needs (CSHCN) programs performed this role.

In 1985, the Council of Regional Networks for Genetic Services (CORN) was developed in response to the need for an organization to facilitate state genetic program efforts through coordination and special initiatives. The CORN published newborn screening system guidelines that defined a 5-part system of screening, follow-up, diagnosis, treatment/management, and evaluation.^{7,9} These guidelines were not treatment guidelines or standards of care, but provided public health agencies with a detailed framework for a systems approach to newborn screening.

By 1985, 12 states had laws allowing charges or fees for screening tests.¹⁸ Today, a majority of the states have established newborn screening fees to be collected from the health care professional, birthing facility, third-party payer, or the parent of the newborn (see Section V, The Economics of Screening). Although newborn screening fees are collected in most states, financing the treatment of children identified with genetic conditions through newborn screening remains problematic. Eligible families in many states are ensured access to therapy (eg, low phenylalanine diet for PKU),^{15,16} particularly when the special formula is deemed a prescription drug. Families deemed ineligible financially may be burdened by the cost of necessary treatments. However, when special PKU formula is classified as a food, many health insurers refuse to cover it at all; creating a problem for both eligible and ineligible families.

Now, after >30 years of experience with PKU, it is clear that knowledge regarding PKU and the approach to newborn screening were rudimentary when the programs were first launched. Studies to validate the screening test, and to assess the safety and effectiveness of a special diet to prevent mental retardation, were completed after laws were implemented. However, the history of these efforts has set the context for the role of public health in newborn screening and genetics.

Setting the Framework for State Newborn Screening Systems

Guidance for newborn screening systems have been in place for 2 decades. These guidelines are inextricably linked to ethical, legal, and social considerations and based on the premise that screening should be conducted only when science and technology can serve both the individual and public good.

Three landmark reports emphasize the criteria that should be used to justify population-based newborn screening systems, and include: the National Academy of Sciences' (NAS) *Genetic Screening: Programs, Principles, and Research* in 1975²⁰; the Institute of Medicine (IOM) report, *Assessing Genetic Risks: Implications for Health and Social Policy* in 1994²⁸; and *Promoting Safe and Effective Genetic Testing in the United States: Final Report of the Task Force on Genetic Testing*, in 1997.²⁹

The NAS Report

The 1975 NAS report set forth rigorous guidelines about the criteria for newborn screening including: evidence of substantial public benefit and acceptance (including acceptance by health care professionals); previous feasibility study; satisfactory test methods; appropriate laboratory facilities and quality control; resources for counseling, treatment, and follow-up; acceptable costs; effective education; informed consent; and the means to evaluate the effectiveness and success of each step. The National Research Council raised concerns about what it saw as the potential risks of inappropriate newborn screening. The NAS report was critical of how PKU screening had developed and suggested the establishment of patient advisory committees made up of individuals with medical and nonmedical expertise.²⁰

The IOM Report

The 1994 IOM Committee on Assessing Genetic Risks recommended that:

"Newborn screening only take place 1) for conditions for which there are indications of clear benefit to the newborn, 2) when a system is in place for confirmatory diagnosis, and 3) when treatment and follow-up are available for affected newborns. . .

The Committee believes that mandatory offering of established tests (eg, PKU, congenital hypothyroidism) that lead to the diagnosis of a treatable condition, is appropriate. If there is no other way to ensure that affected newborns will be identified and have access to effective treatment (eg, in PKU, congenital hypothyroidism), then mandatory newborn screening is acceptable. . .

*Mandatory newborn screening should only be undertaken if there is strong evidence of benefit to the newborn from effective treatment at the earliest possible age (eg, PKU and congenital hypothyroidism)."*²⁸

Although the Committee did point to the appropriateness of the "mandatory offering" of newborn screening tests, they emphasized the use of the informed consent process to educate parents. The IOM report also pointed out that even in cases where a treatment is available for a disorder detectable through newborn screening, timing may or may not be crucial; that is, it may provide no greater or lesser benefit if started after symptoms appear. For example, treatment of children identified through screening for maple syrup urine disease may have only limited effectiveness at best, and parents may face a quandary about whether or not to treat. Even if hypothetical benefits exist, newborn screening systems need close scrutiny to determine if the necessary treatments are actually provided to the children. In states that support screening but not treatment, families may be unable to afford treatment and thus, children may not benefit from screening. For exam-

ple, many children with sickle cell anemia do not get their necessary penicillin prophylaxis and comprehensive medical care. Also, parents of children with PKU are given educational information about diet and nutrition in most states, but not all states provide funds for the expensive essential diet or other food assistance.⁶

The Final Report of the Task Force on Genetic Testing

The 1997 report entitled *Promoting Safe and Effective Genetic Testing in the United States: Final Report of the Task Force on Genetic Testing* pointed out that newborn screening should be of primary benefit to the infant identified. Like the IOM report, the Task Force on Genetic Testing report stated that it would be inappropriate to use traditional newborn screening solely to determine the carrier status of the infant. Moreover, the test should have analytical and clinical validity and utility. Interventions to improve the outcomes for an infant must be safe and effective.

The Final Report of the Task Force on Genetic Testing differed from the IOM Report in that, the Task Force felt that informed consent for newborn screening could be waived, provided that "the analytical and clinical validity and utility of the test" had been established. If the validity and utility of the tests were not established, then informed consent would be required.²⁹

New Challenges Facing Today's State Newborn Screening Systems

As a population-based public health activity, newborn screening systems are housed in state public health agencies. They operate under policies determined at the state level, and ideally, within the framework of the public health core functions of assessment, assurance, and policy development. States vary in public health infrastructure, newborn screening policy establishment, laboratory capacity, screening techniques, as well as in the laws that define the scope of services mandated in response to the identification of a condition. State newborn screening systems also vary in available system components, and in financing mechanisms to pay for these components.

Notably, the array of screening tests performed by each state varies and changes periodically. All state programs now include screening tests for PKU and congenital hypothyroidism. More than 40 programs screen for sickle cell disease and 48 screen for galactosemia. Some newborn screening systems include tests for congenital adrenal hyperplasia, homocystinuria, maple syrup urine disease, and biotinidase deficiency^{6,30} (see Fig 1 and Table 1). A few states also include screening tests for cystic fibrosis, tyrosinemia, additional metabolic conditions, and/or other conditions such as congenital infections (ie, HIV). Over half of the states now require all newborns be screened for hearing loss.³¹⁻³³

The mechanism for deciding which screening tests to include as part of a population-based newborn screening system varies among programs. Thus, the disorders screened for vary from state to state.^{6,18,35-37} These inconsistencies reflect differences

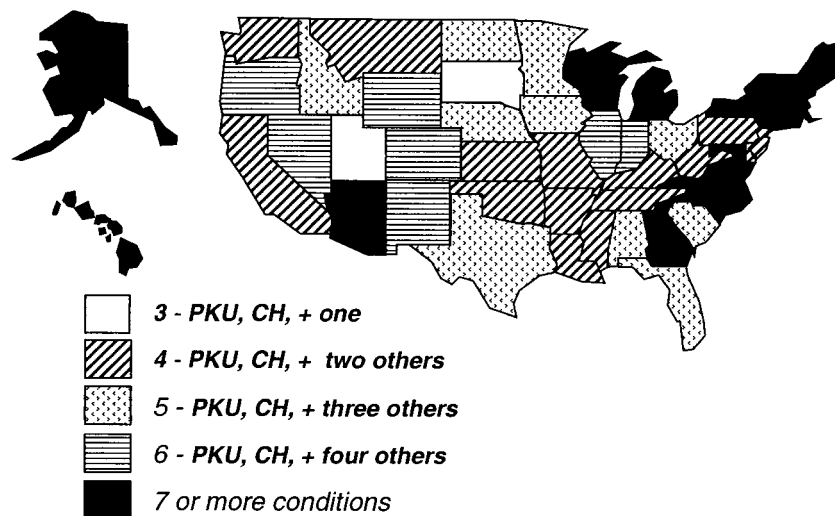


Fig 1. Conditions included in state newborn screening programs, 2000.

in community values, in state political and economic environments, and in public health technical capabilities. Inequities regarding the selection of disorders for newborn screening panels are illustrated by sickle cell disease, a condition for which neonatal screening markedly reduces morbidity and mortality during early childhood. Nationally, sickle cell disease is the most prevalent condition included in newborn screening programs; however, disease prevalence within states varies more than 50-fold because of the widely differing ethnic populations of states. Currently, 41 states and the District of Columbia conduct universal screening for sickle cell disease. Three states conduct screening in infants of high-risk ethnic groups, and 6 states conduct no routine screening. These 9 states are among those with the lowest prevalence of sickle cell disease. Concerns about prevalence, cost-effectiveness, as well as concerns about the acceptability of screening to health professionals and the general public, have hindered implementation of this test despite an NIH-consensus conference recommendation for universal screening.^{3,38-41} In some cases, misperceptions about the benefits of screening, misperceptions about the prevalence of the disease in various ethnic groups, and/or the lack of effective advocacy for the disease have also contributed.⁴²⁻⁴⁵ Thus, while an African-American infant born in a state that does not universally screen for sickle cell disease has the same risk for sickle cell disease as an African-American infant born in a state with universal screening for sickle cell disease, the infant born in the non-screening state is denied the important benefit of screening. In this regard, it is interesting to note that the relatively low prevalence of sickle cell disease in each of the 9 states without universal screening for the disorder (estimated to be >1:40 000) is still higher than the prevalence of galactosemia (estimated to be about 1:60 000-80 000), a disorder included in the screening panels of all 9 states.^{30,46-48} This situation highlights the need for a more uniform national policy for the selection of newborn screening tests.⁴⁹

Because advances in science and technology are

continually making it possible to screen for additional conditions, the decision about which tests to include in a newborn screening panel are complex.⁵⁰⁻⁵⁴ Moreover, in an era of accountability, decision-making is hampered by the lack of studies of and data about test validity and health outcomes. With existing variations between state newborn screening systems, a national model of the structure and function of newborn screening systems has not yet been embraced. Furthermore, there are no uniform guidelines for the periodic assessment of conditions for which screening is performed.^{7,9} As a result, infants across the country do not have equal access to newborn screening and its potential to prevent morbidity and mortality. The US Surgeon General, Dr David Satcher, has emphasized the need for the nation to address unequal access to health care, and the health disparities created by these inequalities. National standards are needed to promote greater comparability of newborn screening programs and address such inequities.⁵⁵

The work of David Hall and his colleagues in the United Kingdom provides useful guidance about creating equitable, sustainable, and effective newborn screening programs.^{56,57} Speaking in Washington, DC, on May 10, 1999, Dr Hall reminded the Task Force of the responsibility to do more than provide screening tests, saying: "If it is important enough to screen for, it is important enough to follow-up." He also spoke on the issues of quality assurance and adequate funding for newborn screening systems stating, "The balance is fine between good and harm in screening. Unless a screening program is a good one, it can do more harm than good."

In the United States, technological advances have had, and will continue to have a significant impact on the sensitivity, specificity, and scope of newborn screening. Pressure is mounting to deploy new diagnostic capabilities despite possessing limited knowledge of their risk and benefit, or their analytical or clinical validity and utility. Presently, tandem mass spectrometry offers, and shortly, DNA-based technology will offer the possibility of using one test or

Table 1. U.S. National Screening Status Report , July 2000

The National Screening Status Report lists the status of newborn screening in the United States. All infants in a state must be screened in order for a dot to be added.

	Phenylketonuria	Congenital Hypothyroidism	Galactosemia	Maple Syrup Urine Disease	Homocystinuria	Biotinidase	Sickle Cell Disease	Congenital Adrenal Hyperplasia	Other
Alabama	•	•	•				•	•	
Alaska	•	•	•	•		•	a	•	
Arizona	•	•	•	•	•	•	•	a	
Arkansas	•	•	•				•		6
California	•	•	•				•		6
Colorado	•	•	•			•	•	a	2, 6
Connecticut	•	•	•	•	•	•	•		2a, 6
Delaware	•	•	•				•		
D.C.	•	•	•	•	•		•		7
Florida	•	•	•				•	•	6
Georgia	•	•	•	•	•		•	•	3
Hawaii	•	•	•	•		•	•	•	6
Idaho	•	•	•	•		•			
Illinois	•	•	•			•	•	•	6
Indiana	•	•	•	•	•		•		6
Iowa	•	•	•				•	•	
Kansas	•	•	•				•		6
Kentucky	•	•	•				•		6
Louisiana	•	•	•			•	•		6
Maine	•	•	•		•	•	a	•	5, 6
Maryland	•	•	•	•	•	•	•		3, 6
Massachusetts	•	•	•	•	•	•	•	•	1,2a,5, 6
Michigan	•	•	•	•		•	•	•	
Minnesota	•	•	•				•	•	
Mississippi	•	•	•				•		6
Missouri	•	•	•				•		6
Montana	•	•	•				a		2a
Nebraska	•	•	•			•	•		6
Nevada	•	•	•	•		•	•		
New Hampshire	•	•	•	•	•		a		1, 6
New Jersey	•	•	•				•		
New Mexico	•	•	•			•	•	•	
New York	•	•	•	•	•	•	•		4, 6
North Carolina	•	•	•				•	•	5
North Dakota	•	•	•					•	
Ohio	•	•	•		•		•		
Oklahoma	•	•	•				•		6
Oregon	•	•	•	•		•	•		6
Pennsylvania	•	•	•	•			•		
Rhode Island	•	•	•	•	•	•	•	•	6
South Carolina	•	•	•				•	•	
South Dakota	•	•	•						
Tennessee	•	•	•	•			•	a	
Texas	•	•	•				•	•	6
Utah	•	•	•						6
Vermont	•	•	•	•	•	•	•		
Virginia	•	•	•	•	•	•	•		6
Washington	•	•	•				•	•	
West Virginia	•	•	•				a		6
Wisconsin	•	•	•	a		•	•	•	2,5
Wyoming	•	•	•			•	•		2, 6

a = Selected population, pilot program, or planning underway

1 = Toxoplasmosis

2 = Cystic fibrosis

3 = Tyrosinemia

4 = HIV

5 = MCAD

6 = Mandatory universal hearing screening

7 = G6PD

simpler tests to detect a larger group of genetic conditions.⁵⁸⁻⁶¹ Furthermore, as the Human Genome Project is completed, the impetus and opportunity to translate genetic knowledge and technology into public health practice will increase.⁶² With these new

technologies comes the ability to detect individuals affected by genetic conditions for which there is no clear advantage to early testing, no early or effective treatment, or no available treatment.⁶³ How should we best use these emerging diagnostic capabilities in

our newborn screening systems and, more generally, in improving the health outcomes of our children?

An updated, consistent national agenda is needed to ensure that state-based newborn screening systems understand and keep pace with new technology. State policymakers and program managers cannot be expected to make optimal decisions in isolation. The process of setting a national agenda for state newborn screening systems requires the involvement of experts in science, medicine, public health, law and ethics, as well as the public and government officials from the federal, state, and local level. The process for these deliberations must take into account public concerns about privacy, confidentiality and discrimination, recent changes in the public health and health care delivery systems, the impact of new advances in science and technology, and the potential cost-effectiveness of revised policies and programs. Such a national agenda can serve as a guide for states seeking to strengthen their newborn screening systems, and provide more equitable access to this public health preventive program for our neonates.

The Task Force on Newborn Screening

To address these and other issues, a national Task Force on Newborn Screening was convened by the AAP, with funding from and at the request of the MCHB, HRSA, and the US Department of Health and Human Services (HHS). Co-sponsors of this effort were: other HHS agencies, the National Institutes of Health (NIH), the CDC, and the Agency for Healthcare Research and Quality (AHRQ); the Genetic Alliance, a consortium of consumer groups; and national public health organizations including the Association of State and Territorial Health Officials, the Association of Maternal and Child Health Programs, and the Association of Public Health Laboratories (APHL).

The AAP was asked to convene the Task Force in recognition that pediatricians and other primary care health professionals must take a lead in partnering with public health organizations to examine the many issues that have arisen around the state newborn screening programs. To ensure that children who are screened are linked to a medical home, it was essential that pediatricians and other primary care health professionals be involved. The AAP defines the medical home as care that is accessible, family-centered, continuous, comprehensive, coordinated, compassionate, and culturally competent. A child who has a medical home, has a pediatrician or other primary care health professional who is working in partnership with the child's family to ensure that all medical, nonmedical, psychosocial, and educational needs of the child and family are met in the local community.⁶⁴

Task Force members were appointed to represent many perspectives and interests among those who operate programs, conduct research, and are affected by newborn screening (See credits page). This report has been approved by the AAP Board of Directors. It does not necessarily reflect the sponsoring organizations' viewpoints, nor do the sponsoring organiza-

tions that provided support for the Task Force necessarily endorse all of the recommendations of the Task Force.

The purpose of the Task Force was to review issues and challenges for these newborn screening programs. The review process was structured to further expand representation. Task Force members were divided into 5 work groups, and additional individuals were invited to participate in each work group's examination of key issues. The work groups were:

- Newborn Screening and Its Role in Public Health,
- Medical Home and Systems of Care,
- Economics of Screening,
- Ethical, Legal, and Social Issues, and
- Implementation and Assessment Issues.

Over the course of 6 months, questions, concerns, and issues were collected from state public health agencies, state public health laboratory directors, MCH programs, pediatricians and other health professionals, families and other consumers, bioethicists, scientists, and health services researchers. Each work group formulated conclusions and developed consensus recommendations. On May 10–11, 1999, the Task Force heard presentations from the 5 work groups, along with public comment on the reports and recommendations. A set of recommendations was developed incorporating key elements of the work group reports, issues raised by the public, and other related information.

Principles and Underlying Assumptions Used to Develop the Task Force Recommendations

Through the past 37 years of experience with newborn screening in the United States and around the world, certain underlying principles and criteria have become widely accepted. The Task Force recommendations are based on the following principles and underlying assumptions.

- Infants should benefit from and be protected by newborn screening systems.
- Not all conditions are good candidates for newborn screening. The criteria for inclusion of a screening test are: a) the condition is an important health problem that occurs frequently enough to justify screening an entire population; b) the treatment for the condition is effective when initiated early, accepted among health care professionals, and available to all screened newborns; and c) the test is simple, safe, precise, validated, and acceptable.
- Newborn screening is more than testing—it should always be part of a system that includes screening tests, follow-up, diagnosis, treatment, and evaluation as necessary. The primary objective of each state's newborn screening system should be to ensure that every newborn receives appropriate and timely services.
- Newborn screening is an essential public health prevention activity that requires integration of parent education, sample collection, laboratory analysis, primary and specialty medical care, and related services for families with affected children.

- State public health agencies should assume responsibility for assessment, assurance, and policy development in the context of newborn screening, giving particular attention to the adequacy of system structures, oversight, and funding.
- The complete newborn screening system (testing, follow-up, diagnostic procedures, treatment, and evaluation) should be clinically, socially, and ethically acceptable to the public and health professionals.
- Infants should have a “medical home” (identified by parents before or after birth) that is linked to a newborn screening system and includes access to appropriate care and treatment, if a condition is diagnosed.
- Infants born anywhere in the United States should have access to screening tests and procedures that meet accepted national standards and guidelines. New screening tests should meet national criteria for newborn screening, with data on the validity of new tests and the clinical utility of screening new diseases collected through pilot programs.
- Before newborn screening, parents (on behalf of their children) have a right to be informed about screening, and have the right to refuse screening. They also have a right to confidentiality and privacy protections for information contained in all newborn screening results.
- Increased coordination and uniformity, among state newborn screening systems and other child health programs, will greatly benefit families, health care professionals, and public health agencies.
- Parents and consumers must be involved in all parts of the policy-making and implementation process.

Screening and Counseling for Genetic Conditions in 1983

The President’s Commission for the Study of Ethical Problems in Medicine and Biomedical and Behavioral Research made recommendations entitled “*Screening and Counseling for Genetic Conditions*” in 1983.¹⁶ Many of these earlier recommendations have striking resonance today, despite the advances in science, technology, and medical care. The following findings (excerpted from that report) illustrate the continuity between earlier consideration of ethical issues in screening, and the work of the Task Force on Newborn Screening. The Commission found that:

- The parties involved, including regulators, funding agency administrators, industry representatives, researchers, and public health officials, should meet to discuss their respective roles in ensuring that a prospective test is studied adequately before screening programs are introduced.
- Successful programs require concrete goals and specific procedural guidelines that are founded on sound ethical and public policy principles.
- If ethical and policy goals are to be promoted, every screening program should have an evaluation component.
- Oversight bodies can provide an important focus for the successful provision of services.

- Public screening programs should not be implemented until they have first demonstrated their value in well-conducted pilot studies.
- Cost-benefit analysis must be regarded as a technical instrument to be used within an ethical framework, rather than as a method of avoiding difficult ethical judgments.

Task Force Assumptions Regarding the Future of Newborn Screening

The value of a blueprint depends in large part on how well the architects understand the setting. Although many unforeseeable events may change the landscape, an assessment of the environment is essential. The Task Force anticipates that the following trends will affect the future of newborn screening over the next 5 to 10 years.

- The newborn screening system affects many people and institutions, which in turn creates potential for problems related to conflicts and gaps in follow-up and services. With changes in the health care delivery system, financing, medical practice, and public health agency structures, such problems are likely to increase.
- The dramatic advances in genetic science are changing the environment for newborn screening. As the Human Genome Project is completed, the expansion of genetic knowledge and technology into public health will continue, presenting opportunities for understanding and promoting better health, lowering mortality and morbidity, and preventing diseases. As the Human Genome Project reaches fruition, medical genetics and the number of genetic risk factors for diseases that can be detected will grow rapidly. New DNA-based testing technology will be one outgrowth of applied research.
- Technological advances have had and will continue to have a significant impact on the sensitivity, specificity, and scope of newborn screening. Tandem mass spectrometry offers the possibility of detecting a larger group of metabolic conditions. With this new technology comes the ability to detect individuals with metabolic conditions for which there are no effective treatments at this time.
- With recent and future technological advances, newborn screening can also be used for more than testing for hematologic, endocrine, metabolic, and other genetic conditions. For example, newborn hearing screening is a widespread newborn screening procedure in the United States that currently does not use a blood sample or DNA-based testing technology (although hearing loss may be genetic in origin and blood samples may be used for DNA confirmation in the future). The future will bring more opportunities for early screening and systems integration.
- States will continue to be the policy innovators and primary regulators for health care, including insurance, public health, patient rights, and professional and facility licensure.

- Partnerships and collaborations between medicine and public health will be expanded and better developed. For newborn screening, immunization, and other services that operate at the intersection of clinical medicine and public health, effective collaboration is essential to achieving positive outcomes.
- Newborn screening will operate in a health care marketplace that depends on public-private ventures. State agencies will further expand private sector purchases and contracts for health-related services.
- The demand for consumer protections in health care will continue to be high; including demands for privacy and confidentiality protections of medical records and health information. These demands, along with parental opinions, will influence the future of newborn screening.
- Public perception of genetics in medicine and technology will lag behind scientific advances. Even among the better-educated general public, the perception of risks and benefits may differ from the views of health care professionals.
- Policymakers will continue to respond to concerns about the health of children and show some willingness to make investments in child health protections and prevention tools such as screening.
- Broader child health issues will continue to influence newborn screening systems; as newborn screening is only one part of child health surveillance, and infants with identified conditions are only one set of CSHCN.
- Health care cost containment pressures will continue to have substantial influence in health policy. If premiums for health coverage continue to rise, purchasers (eg, employers and government) and policymakers will take action. The response may lead to reduced coverage for new tests and treatments, greater inequities based on income level, and/or greater numbers of uninsured individuals and families. The result will be continued reduction in the quality of care.
- Health professionals will require ongoing training and education in newborn screening and new technologies. Additionally, pediatricians and other primary care health professionals who care for children should receive training on their role as the source of a child's medical home.

Advancing a National Agenda for Newborn Screening

The CORN guidelines^{7,9} and recommendations from previous expert panels and task forces form a foundation for advancing newborn screening. Despite some areas of disagreement (particularly on the topic of informed consent and parental permission), these documents together outline similar principles for conducting newborn screening (eg, the condition is serious, early screening would benefit infants, a reliable test is available, treatment is available, and early diagnosis and treatment are important to the infant). However, even when there is consensus, some state newborn screening systems have not applied these recommended standards and guidelines when setting policy and program structures.

In recommending model regulations and national standards, the Task Force recognizes that the translation of any models would have to conform to a state's particular infrastructure and infrastructure needs. The Task Force believes that public health agencies (federal and state), in partnership with health professionals and consumers, should continue a process that will:

- Better define public health responsibilities for federal and state public health agencies;
- Develop and disseminate model state regulations to guide implementation of state newborn screening systems (including disease and test selection criteria);
- Develop and evaluate innovative testing technologies;
- Design and apply minimum standards for newborn screening activities (eg, sample collection, laboratory quality, sample storage, and information systems);
- Develop and disseminate model follow-up, diagnosis and treatment guidelines, and protocols for health professionals and other participants in the newborn screening system;
- Design and evaluate model systems of care with services and supports from infancy to adulthood that are consistent with national guidelines for CSHCN (ie, family-centered, community-based, and coordinated systems of care);
- Design and evaluate tools and strategies to inform families and the general public more effectively; and
- Fund demonstration projects to evaluate technology, quality assurance, and health outcomes.

The Task Force has made further recommendations to address specific concerns and has identified needs for program and policy development in 4 key areas: Public Health Infrastructure; Professional and Consumer Involvement; Surveillance and Research; and The Economics of Screening. (Each topic is discussed extensively in later sections of this report.)

By outlining these recommendations, the Task Force seeks to further advance consensus. The Task Force recommendations call for change in many facets of state-based newborn screening systems. This work is intended to inform policy decision-makers about the possible strategies for enhancing newborn screening systems. The Secretary's Advisory Committee on Genetic Testing (SACGT) is expected to develop recommendations for the US Secretary of HHS regarding the oversight of genetic tests with respect to the accuracy, meaningfulness, and appropriate use. Newborn screening is among the issues the SACGT is addressing. Infant hearing screening, and other types of newborn screening, deserve similar attention from the federal and state policy communities. State legislators and executives face the challenge of deciding what tests, what testing technology, and what resources to use in protecting their pediatric populations.

Parents have served as advocates to advance newborn screening policy since the 1960s, and this con-

tinues to be an important role for them. Also, pediatricians and other primary care health professionals who care for children must participate in the development of guidelines for practice and policy. Joint leadership from government, health professionals, and parents will be essential if a nationwide approach to newborn screening is to be designed for the future, and if changes are to be implemented in each state.

Success of Newborn Screening

Newborn screening has been one of the nation's most impressive recent public health achievements and one of the most reliable components of child health services. The Task Force reaffirms that our nation's programs for newborn screening have improved the health and well-being of our children. The Task Force recommendations for newborn screening call for changes in many facets of these state-based systems, because the achievements of the past are not sufficient to carry newborn screening systems into the 21st century. Much has changed since most newborn screening systems were de-

signed 30 to 40 years ago. Success in the future will depend on the adequacy of our response to new genetic science, advances in knowledge about infant development, evolving biomedical technology, and changes in health care delivery and financing. Strengthening the newborn screening systems, as laid out in this blueprint, will require attention to the need for an improved public health infrastructure, the gaps in public and professional involvement, the challenging research agenda, and adequate financing. The intellectual and fiscal resources needed to achieve continued success are within our means and can be dedicated to the tasks ahead if there is political will to do so. Leadership from government, health professionals, and parents will be equally important to craft a national agenda for newborn screening and to implement changes in each state. Newborn screening can lead to early identification and treatment of about a dozen conditions today and perhaps scores of conditions by the year 2010. Well-functioning newborn screening systems are important to the 4 million US children born each year, and deserve the nation's attention.

II. PUBLIC HEALTH INFRASTRUCTURE

Newborn screening systems must be placed within an adequate public health infrastructure, since newborn screening involves more than testing. A screening test will be effective only if it is placed within an appropriate infrastructure that includes: education for the consumer and public, sample collection, laboratory tests, follow-up, diagnosis, appropriate treatment, information management, and system evaluation. The primary objective of each state's newborn screening system is to ensure that every newborn receives appropriate services. Public health agencies, at both the state and federal level, have a responsibility to ensure the quality of the newborn screening system. This section explores the history of newborn screening in public health agencies, the impact of new technologies in the evolution of screening programs, and the roles that can best be played by public health agencies in newborn screening systems.

The History of Newborn Screening as a Public Health Agency Role

Public Health Mission and Core Functions

To define the role of public health in newborn screening, one must understand how the level of scientific and technical knowledge, as well as public values, has changed over the past 37 years.

No single definition of the general role or mission of public health exists. Some describe public health as "population-based" activities that benefit everyone, as with prevention, or others see it as the health care of last resort. Although private organizations and individuals play important roles, governmental public health agencies have a unique function and responsibility to address this mission.

The IOM defined 3 core functions for public health agencies:

- Policy development—the responsibility to serve the public interest by promoting use of scientific knowledge in decision-making and policy development.
- Assurance—the responsibility to ensure that services are provided, either by encouraging action by other entities, requiring action through regulation, subsidizing services, or providing services directly.
- Assessment—the regular and systematic collection and analysis of information on the health of the community.⁶⁵

Public health programs and operations have conducted these roles since the 1890s. The first state and local health department laboratories concentrated on improving sanitation. Registries were established to track the spread of infectious diseases. Since that time, the role of detecting disease has continued to be an important one. Public health agencies were able to carry out activities that affected individual rights, such as these, because they operated under the authority of the state to protect the public's health.

Public health agencies have also been used throughout the 20th century as a base for diverse

activities including: home visits by public health nurses, health education, treatment of tuberculosis, childhood immunizations, as well as to directly furnish a broad package of clinical services. Between 1970 and 1990, many agencies used available resources to subsidize or provide services directly to medically indigent individuals, while maintaining the other core functions and responsibilities. During the 1990s, Medicaid was expanded to provide health coverage for low-income individuals, and was "privatized" through contracts with managed care plans.

Newborn Screening Becomes a Public Health Agency Role

With the advent of tests for PKU and other metabolic conditions in the 1960s, the responsibility to implement newborn screening systems was assumed by state public health agencies.^{12,17,18} Only public health agencies—using their authority to protect the public's health—could implement systems that would assess the prevalence of conditions, mandate newborn screening for all infants, ensure the quality and availability of testing, and provide follow-up on a population basis. Today, with new genetic technology and changing public opinions about the role of the government, the public health agencies' role in newborn screening systems is evolving.^{66–69}

Challenges Facing Today's Public Health Agencies

State health agencies of the 1990s are different from those of the 1970s.^{65,70} They face the challenges of keeping up with new testing technology, responding to emerging infections and a resurgence of old diseases (ie, tuberculosis), coping with budget cutbacks or windfalls (eg, tobacco legislation funds), and operating in a new health care delivery system (eg, managed care arrangements, integrated health systems).^{71–73} At the same time, state public health agencies remain charged with assessment, policy development, and assurance functions.

Challenges Related to Newborn Screening

Some challenges most directly related to newborn screening include:

- Laboratory capacity is sometimes inadequate. The methods used by some health department laboratories are in need of enhancement. In addition, public health laboratories are often under competitive pressure from commercial laboratories.⁷⁴
- Budget constraints make it more difficult for health departments to cope with the current workload, let alone with new tests that require additional equipment and personnel.
- Benefits of some screening tests have not been appropriately validated.
- State public health agencies screen for different conditions. These differences are not always based on the prevalence of the disorders in the respective states, or proof of the tests' utility and validity.
- Funding is insufficient for newborn screening quality assurance and evaluation, particularly for laboratory and database information systems.
- State policymakers possess an incomplete understanding of the conditions for which newborn

screening can be conducted, and/or of testing technology.

- Informing and educating consumers is often challenging, and meaningful public and consumer involvement is not always considered.
- Adequate funding is needed for comprehensive care by multidisciplinary teams in medical homes, including resources that ensure the availability of special formula, special foods, and other treatments for all affected children and adults.

Lack of Uniform Public Health Policy on Newborn Screening

Since newborn screening began, all states and territories of the United States have included newborn screening as part of their preventive public health system.^{75,76} Considerable variability exists in: the systems available for follow-up, the genetic conditions included as part of the screening system, the laboratory capability within the state, the treatment protocols, and the scope of follow-up services mandated as part of the newborn screening system.^{6,7,18,27,38,77} This situation highlights the need for uniform national policy on the selection of newborn screening tests, as well as common guidelines for newborn screening systems. Without nationally observed standards, infants across the country do not have equitable access to newborn screening, and its potential benefits. State legislators, health commissioners, and newborn screening system managers benefit from nationally recognized standards and guidelines by having recognized and well-considered benchmarks for the development of their programs.

The Task Force's Response to These Challenges

A continued role for state health departments in management, coordination, and evaluation of newborn screening programs is vital to sustaining and improving newborn screening systems. The Task Force concludes that a public health role is essential to continue newborn screening programs at their current levels. In facing current and future challenges, states and their public health agencies need to address the following questions:

- What steps are necessary to assist state policymakers in making decisions about tests and testing technology?
- How could a national, minimum set of newborn screening tests and standards be developed for use by states and their public health agencies?
- How can quality assurance and evaluation be better financed and utilized by public health agencies?
- How can public health agencies best carry out their quality assurance responsibilities in conjunction with private sector health care professionals, laboratories, and other entities?
- How could data and information efforts be improved to ensure the follow-up, tracking, and evaluation needs of newborn screening systems?
- What is the role of public health agencies in coordinating these efforts to ensure that they serve

families in the most efficient and effective manner?

- How can public health agencies ensure that the pediatrician or primary care health professional who is the source of the child's medical home receives newborn screening results in a timely and efficient fashion, even when the results are negative?
- How can public health agencies play a more active role in long-term management and follow-up from infancy to adulthood?

Designing and Developing Newborn Screening Systems

Decisions Regarding Tests and Testing Technology

In all states today, every infant is screened for 2 disorders: PKU and congenital hypothyroidism.^{6,7,9,18,30,48} Beyond these 2 tests, there is inconsistency between states in the panel of conditions screened. In addition, new advances in science and technology are continually making it possible to screen for additional conditions. The decision about which tests to include in a newborn screening panel is becoming increasingly complex. As a result, one role of the public health agency is to ensure that adequate data are available to decide whether a screening test should be included in the repertoire of routine tests.

If tests are outdated and need modification, or if the state public health agency feels that tests should be added or removed, challenges may exist in implementing these changes.^{78,79} A number of state programs offer tests prescribed by law, and must seek legislative change before making program change. In this case, it is preferable to seek legislative authority to allow program change through the rule-making (ie, regulatory) process. Many programs already have laws allowing program changes by rule (regulation).^{6,18,23} To add structure to such program change, it is preferable to adopt guidance for such considerations and debates. In accordance with the CORN guidelines for newborn screening systems,⁷ newborn screening program guidance in each state should include defined parameters such as:

- Demonstrated value to the affected patient and to the public through screening, detection, diagnosis, and treatment in a pilot program;
- A publicly accepted mechanism for funding the change which ensures that screening, follow-up, diagnosis, and treatment services will be available even if the family is unable to pay;
- Demonstrated cost utility, showing benefit in quality-adjusted years of life and reduced public health impact (prevalence × severity × effectiveness of intervention = public health impact);
- A mechanism for evaluating and ensuring quality throughout all elements of the screening system^{80,81}; and
- A system for educating all stakeholders as to the benefits of the program and its changes.

The Human Genetic Society of Australasia also defined parameters for inclusion or exclusion of con-

ditions screened for newborn screening programs.⁸² These categories include:

- Recommended for screening (a demonstrated benefit from early diagnosis exists that is balanced against financial and other costs, and for which suitable tests and follow-up services exist);
- Recommended if resources permit (a demonstrated benefit from early diagnosis exists that may not be balanced against financial and other costs depending on the available technology, the frequency of the disorder in the region, and other local circumstances);
- Pilot-screening recommended (benefit to the individual from early diagnosis appears likely, it is likely to be balanced against financial and other costs if suitable technology is available, there are tests available that are likely to be suitable, and there are follow-up services available);
- Screening tests are available but not currently recommended (proof of advantage from early diagnosis is absent or uncertain or the test is unsuitable or does not detect those cases in which there might be an advantage); and
- Conditions that may be detected incidental to screening for a recommended disorder (properly constituted research programs into the utility of screening for the disorder is encouraged).

Developing Adequate Follow-up Systems

Deciding which screening tests to include is just one aspect of the newborn screening system. To attain the greatest possible benefits of newborn screening, careful follow-up and continuity of care must also be ensured.^{7,9}

The role of state and local public health agencies in the initial follow-up of newborn screening varies widely.^{6,19,77,83–89} Some states assign the laboratory (public or private) the responsibility to communicate results to the health professional or facility that will follow-up with families. Other states provide active support by using local health department staff (usually from Title V MCH programs) to identify the medical home, locate the family, or communicate test results. When a child's family cannot be readily located, follow-up through mail, telephone, or direct contact through home visits may be necessary to ensure that a diagnostic test is done and treatment is initiated if warranted. Other state-financed follow-up activities may include public health nurses to collect blood specimens, nutritionists to help families establish and maintain dietary control for their children, and social workers to give support to families of affected children.

If an infant is identified and confirmed as having a specific disorder, follow-up with pediatric subspecialists and pediatric subspecialty clinics is often needed. In some cases, networking relationships between pediatric subspecialists and the infant's pediatrician/primary care health professional already exist. For example, some states simultaneously share sickle cell disease, congenital hypothyroidism, and PKU test results with the infant's primary care health professional and a pediatric subspecialist (eg, hema-

tologist, endocrinologist, or geneticist).⁴⁷ Health care professionals report that such links, through the newborn screening system, can simplify the follow-up process.⁴⁷

Follow-up of newborn screening tests is an activity that is performed by many health departments today.^{11,25,65,88,90,91} Most consider one of their roles to be providing or "enabling" support services (eg, care coordination, transportation, and information). This enabling role, that assists families seeking services and other public health agencies, is a base for home visiting programs. Evidence indicates that families and health professionals welcome this type of support. Generally commercial laboratories have not provided the same type of support services and assurance function essential to the newborn screening system.

Developing Newborn Screening Systems Through Education and Collaboration

Another public health agency role is to increase awareness of newborn screening among health professionals, parents, and the public. The success of newborn screening systems depends on the knowledge and behaviors of these individuals. An improved understanding of newborn screening and genetic medicine, and the benefits of the newborn screening public health program are essential.

In addition to providing education to health professionals, parents, and the public, collaboration among these groups, facilitated by public health agencies, is crucial. Multidisciplinary participation in newborn screening program advisory boards is one way that this collaboration can take place. The seamless integration and thoughtful collaboration among these participants is of vital importance to the smooth functioning of a universal newborn screening program. Partnerships must be maintained, so that the system's effectiveness can be sustained. (For further discussion, see Section III: Professional and Public Involvement.)

Quality Assurance and Evaluation

Ensuring the Quality of Newborn Screening Laboratories

Laboratories performing testing, in the public interest, are generally driven by 2 principal factors: cost-efficiency and quality. Ideally, newborn screening testing is inexpensive, produces high-quality results, and is technically advanced. In reality, it is often difficult to balance all of these factors within the political and economic environment of a state and a public health program.^{7,14,29} Therefore, it is incumbent on all programs to monitor laboratory performance and technological progress. It is thought that to maintain optimal quality, sufficient positive testing results should be encountered so that a positive test is easily recognized. There is no universally accepted standard in this regard, and high-quality laboratories exist with both low and high volumes of testing. In newborn screening, it has been recommended that the threshold number of samples should be 30 000 annually.⁷

In almost all state and territorial newborn screen-

ing systems, a public health laboratory provides testing.^{6,14} One potential problem is a low volume of cases and related cost and quality issues.⁹² In these cases, solutions can be sought jointly between the program and the laboratory. Some programs have found that laboratory regionalization and laboratory contracting offer possible solutions to this dilemma. Regional laboratories exist where states have agreed to pool their testing volume into a single laboratory, to maximize economies of scale.^{26,93} Other states use contractual arrangements with private or public laboratories. This approach may reduce costs or provide additional capacity not otherwise available. In either case, it is the responsibility of the state health agency and its newborn screening system to ensure the highest quality laboratory services for its constituents through laboratory monitoring and quality assurance procedures.

Today, all newborn screening testing must be performed by laboratories that meet the requirements of the Clinical Laboratory Improvement Amendments of 1988 (CLIA '88), which include criteria for quality control and proficiency testing programs.⁹⁴ Proficiency testing is a tool used to evaluate the quality of a laboratory's testing process. This involves a monitoring organization sending proficiency testing specimens to laboratories on a periodic basis, usually quarterly. Proficiency testing specimens are then handled and analyzed in the same manner as patient specimens; with results sent back to the monitoring organization for evaluation. This testing helps to ensure the quality of each laboratory's measurement process. Laboratories must satisfactorily participate in a Health Care Financing Administration (HCFA)-approved proficiency testing program, if available, for each laboratory method they use to analyze human specimens.

Special expertise in dried blood spot technology is required in both newborn screening testing and quality assurance. Further, because quality assurance services would be provided to a small number of public health laboratories, it was thought that it would be burdensome for participating state laboratories to provide sufficient fees to support a national quality assurance effort. Therefore, more than 20 years ago, the NAS recommended that a single laboratory within the CDC be responsible for maintaining the proficiency of the regional laboratories conducting newborn screening for metabolic disease.²⁰ The CDC pursued this recommendation when the Genetic Services Branch, MCHB, HRSA offered to help support the development of a national quality assurance program at the CDC, which has come to be known as the Newborn Screening Quality Assurance Program (NSQAP). The NSQAP has enabled laboratories to meet the CLIA quality-assurance requirement for verifying test accuracy. This was particularly important in the absence of an HCFA-approved proficiency training program for newborn screening. This collaborative effort between the HRSA and the CDC (with the cooperation of the APHL) was based on recognition that newborn screening is a major public health effort mandated by laws in most states.

The HRSA's 20-year funding for the CDC's oper-

ation of the NSQAP ended in 1999, based on a recognition that emerging newborn screening technologies, such as DNA-based testing, required the involvement of the SACGT and other HHS agencies—the Food and Drug Administration, the HCFA, the National Institutes of Health, and the CDC—to address the regulatory and research needs related to quality assurance.

In the absence of HRSA funding for the NSQAP, a new mechanism for providing oversight and assuring quality in laboratories nationwide must be developed and funded. Moreover, as new screening technologies and modalities are put into practice, ensuring quality for all children in newborn screening systems depend on such a nationwide effort.

Evaluation of the Newborn Screening System

States play an active role in developing the structuring and financing mechanisms for quality assurance, accountability, and oversight of newborn screening systems. Although many state MCH programs, using their federal Title V Block Grant and state matching funds, play a key role in quality assurance for each of the first 4 components of the newborn screening system (screening, short-term follow-up, diagnosis, and treatment/management), the fifth component, evaluation will need to be addressed.

Public health agencies have a responsibility to evaluate the performance of the newborn screening system. This responsibility was broadly outlined by the IOM in 1987, and legislatively by the Title V Social Security Act for MCH programs.⁹⁵ For example, the new guidelines for state MCH programs (developed in response to Government in Performance and Results Act requirements) set out national performance measures for states. In addition, states have an opportunity to set additional performance objectives based on their needs and priorities, and some states use additional performance measures related to genetic conditions, birth defects, and/or newborn screening.

The CORN guidelines specifically emphasize the importance of evaluation in achieving the goals of newborn screening systems and ensuring that they operate in the most effective, efficient, and cost-effective manner.^{7,9} This component includes process evaluation of the state public health activities, as well as outcomes evaluation of the newborn screening system overall. At a minimum, state health agencies must complete a review and evaluation of their newborn screening activities (internally or externally). This includes quality assurance elements such as review of laboratory quality, appropriateness of specimen storage methods, rates for completion of repeat testing, and rates for completion of follow-up. Furthermore, current HRSA consultative program reviews, through its cooperative agreement to the National Newborn Screening and Genetic Resource Center, should be continued.

Program evaluation and quality assurance mechanisms in newborn screening systems need to be strengthened. On a population level, it is clear that mental retardation attributable to PKU and congen-

ital hypothyroidism, and mortality and morbidity from sickle cell disease in early childhood have been reduced.^{40,96–98} Evidence indicates that neonatal morbidity and mortality from maple syrup urine disease and galactosemia also have decreased.⁹⁶ At the same time, accurate and timely data are not available to measure the proportion of infants screened, proportion of infants with positive tests promptly and adequately followed-up, or success in terms of prevention of disability or other morbidity and mortality. On an individual level, data are not available to determine the range of functioning of affected children at various ages, the relation of function to care received, and the other benefits and risks (eg, parental anxiety, effect on unaffected siblings).

States should maintain ongoing outcome evaluations. State public health agencies play a role in defining performance and outcome measures. For example, under the HRSA Title V MCH Block Grant Program, state health agencies are accountable for reporting on 18 performance measures, including 4 that are directly related to newborn screening (see Section IV: Surveillance and Research). Additional state and local measures for newborn screening systems might focus on outcomes such as survival, and health and functional status; process factors such as time from test to diagnosis, and percent of repeat screens completed; and quality-related factors such as parental involvement and satisfaction, and number lost to follow-up in the course of specialty care.

Economic measures for cost-effectiveness or cost-benefit studies have been used in the past to assess the appropriateness and social utility of newborn screening. Screening for certain disorders, such as PKU and congenital hypothyroidism, have been shown to be cost-effective. However, economic analyses and evaluations must take into account that, while screening may save the lives of some infants, the long-term care costs will sometimes be higher than not screening. Thus, the use of cost-savings as the justification hinges on having a treatment that reduces long-term costs. Caution is warranted in only using economic measures as evaluation tools or outcome indicators (see Section V: The Economics of Screening).^{5,9,99–103}

Federal grants could be used to stimulate newborn screening information systems with an emphasis on outcome measurement and evaluation of effectiveness and cost-effectiveness. Such grants might provide incentives and start-up funding for outcome data collection systems, development of uniform data sets, and similar activities. As the health care system evolves—with the application of genetic medicine, new testing modalities, new delivery systems, and new technological tools to manage data and information—states face substantial challenges and have major opportunities to carry out the role of quality assurance.

Ensuring the Quality of Private Sector Activities Related to Newborn Screening

Private professionals and facilities carry out a number of newborn screening activities. These include specimen collection in hospitals, specimen

transport by private courier, laboratory tests by private laboratories, follow-up by private contractors, diagnosis and treatment at private specialty centers, and research by private institutions.¹³ Where the state health agency has only indirect responsibility, it has legislative and regulatory powers to ensure newborn screening system quality.^{18,19,53,104} This may come in the form of licensure requirements, reporting requirements, public health guidelines, contract specifications, and so forth. State public health agencies must develop collaborative approaches and linkages to private physicians, hospitals, laboratories, and others to ensure optimal coordination.

Integrating and Coordinating Related Programs

A Review of Related Programs

Public health agencies and other government programs have multiple programs designed to serve infants in the first month (neonatal period) or first year of life.^{105,106} The following programs are among those that logically could be connected to newborn heelstick screening programs. Each provides screening for other conditions, includes follow-up and tracking components, or is aimed at serving CSHCN.

Programs screening infants for other health and developmental risks

Prenatal screening and follow-up. Screening tests done prenatally may require follow-up treatment of the newborn infant.^{102,103,107–109}

Newborn hearing screening. An increasing number of states are implementing universal newborn hearing screening, shifting from policies that had previously emphasized hearing screening only for those infants with recognized risk factors for hearing deficits.^{31–33,84,110–114}

Supplemental Nutrition Program for Women, Infants, and Children (WIC). Substantial proportions of pregnant women and newborn infants receive nutrition support through the WIC. (In some states and cities, from 50% to 75% of infants meet income eligibility guidelines for the WIC.) Screening for nutritional risk is a core function of the WIC programs, and some also screen for immunization status and/or development risk.¹¹⁵ Historically, WIC programs have supported formula for management of PKU (ie, Colorado).

Outreach, case management, and home visiting programs. Child health programs that seek to identify children with varying levels of social or medical risk may involve outreach, case management services, or home visiting. In some areas, public health nurses may seek to provide a home visit for a large proportion, or all, infants and families shortly after birth. Such services may be a continuation of support initiated for mothers in the prenatal period.^{116–121}

Programs for infants with or at risk for special health care needs

High-risk infant follow-up programs. Many states provide programs through which children discharged from newborn intensive care units receive

follow-up services and may be enrolled in special developmental follow-up clinics.^{122–124}

Early intervention programs. PL 99-457 and subsequent amendment in the Individuals With Disabilities Education Act led to creation of programs designed to identify and provide services for children birth to age 3 with, or at risk for, potentially disabling conditions. While states may choose among the categories of children to be served, screening for eligible children and development of an “Individualized Family Service Plan” for follow-up and treatment is required in every state. These very young CSHCN may be linked to both health care and related developmental and educational services.^{125–127}

State genetics services programs. Genetics programs may provide evaluation, diagnosis, long-term treatment, and case management for children with genetic disorders, including those identified by population-based heelstick newborn screening and birth defects registries.¹²⁸

Registry and data programs that include infants

Vital registration. Birth certificates are increasingly filed electronically. These electronically filed certificates may serve as the foundation for an electronic health record, including newborn screening status, and might be populated from newborn screening contacts.

Immunization registries. Infant immunization services may begin in the newborn nursery and continue throughout life. With substantial federal support and the involvement of private foundation and corporate resources, states are developing computerized immunization registries, using electronically filed birth certificates as the basis for initiating entry into the registry.¹²⁹

Birth defects registries. Increasingly states use birth defects registries to identify children with congenital abnormalities that require treatment and follow-up, to study the causes of these conditions, and to plan for services.¹³⁰

Challenges Involved in Coordinating Programs and Information Systems

Publicly funded infant and child health programs often operate independently of one another. The resulting duplication of effort can increase costs, burden families and health professionals, and create redundancy in data management systems. Improved coordination and integration of information systems is needed.^{131,132}

State agencies attempting to coordinate infant or other public health programs face a variety of challenges. These programs serving infants each operate with potentially varying time frames (ie, filing electronic birth certificates may take 2 to 3 weeks, but data for newborn heelstick screening needs to be entered within days of birth), definitions of eligibility (ie, universal heelstick screening versus means-defined WIC eligibility), demands and constraints imposed by categorical funding agencies, and priorities. In addition, they may be administered by different agencies within state governments.

As a result, services may not be well-integrated or coordinated. This can lead to the inefficient use of resources and frustration among families who are frequently asked to provide the same information on multiple forms of varying formats or categories. Information systems that support these programs may be insufficient, redundant, or independent of one another. Program integration and coordination cannot be achieved without a substantial new investment in infrastructure, and without addressing complex policy issues such as the confidentiality of health information.⁹⁰

Ironically, one of the unintended effects of this lack of coordination and/or communication among programs and data systems may be a form of greater confidentiality protection; the current system does not allow easy aggregation of personal information. This scenario leads to the following questions: 1) What is the optimal framework for integrating or coordinating public health systems for newborn assessment and follow-up? and 2) What is the role of information systems as part of efforts to improve program coordination? Technically, it is now feasible to link data systems; however, ensuring the proper use of data and adequate privacy protections may be difficult. Parents, health professionals, program managers, and public health officials may each have different goals and perspectives.^{132,133} Thus, in considering whether to integrate programs and their information systems, and how to go about this, it is essential to take into account the benefits, as well as the liabilities and costs to each group.

The value of efforts to link, coordinate, and integrate programs should be measured against the following criteria:

- Is duplication of effort reduced?
- Is the knowledge of resources and services improved?
- Is access to resources and services improved?
- Is the quality of services for children and families improved?
- Can appropriate privacy and confidentiality protections be ensured?
- Is the work of service and health care professionals facilitated?
- Is program management improved?
- Can improvements in public health be documented?
- Is there an improvement in child health?

Family perspective. Priorities from the perspective of families are likely to include:

Access to relevant information about the child. Parents and families are not apt to be concerned about the architecture of information systems. However, they are interested in having the information they need to make informed decisions in the interest of their child’s health.^{134,135} They also desire timely access to various forms required for documentation of need or service, such as documentation of immunizations for school enrollment. In some instances, parents may have to balance their conflicting desires for easy access to services and protection of privacy (ie, a reg-

istry that gives ready access to immunization records requires that parents give permission to store the record).

Services ensured with continuity. A family's primary concern is that access to services is ensured, and that the array of services that children require be provided as seamlessly as possible, with minimal effort required in negotiating the system of care. A family also needs to know what services are available as they make choices for their child.

Privacy protection. The protection of privacy is often a paramount concern among families. Some may object entirely to their child being included in a government-sponsored registry.

Health care professional's perspective. As advocates for their patients, health care professionals will share many of the same interests as their patients and the patients' families. Health care professionals also are likely to have these additional interests^{136,137}:

Timely and ready access to accurate information about their patients. The health care professional wants the relevant screening information about that child readily available. For some professionals, or their staff, whether data are available through electronic systems may be less important than simply being ensured that the information can be obtained. For others, access to electronic child health records would facilitate their work. To the extent that computerized and other electronic systems are being developed as sources for program information, the health care professional (or staff) would prefer to tap into a single system to obtain data from different programs, rather than having to check multiple data systems to obtain information. For example, information from a single point of contact would ideally include: immunization status, and the results of hearing screening and heelstick screening. Easy access to equivocal, positive, or negative results of newborn screening is desirable, but is only one type of information a medical home may need.

Avoiding missed opportunities for follow-up at multiple points of contact with the child. Many children receive services at multiple locations or facilities. Ideally, information would be available to each health care professional, as necessary. Moreover, each professional should be aware of the need for follow-up on newborn screening and other conditions, regardless of the purpose of a particular visit. For example, if one of your colleague's patients is being seen in your office, you should have the ability to identify that the child needs a repeat PKU screening test, or that the child has failed their first hearing screen. This will allow the health care professional to take advantage of the opportunity for follow-up.

Minimizing duplication. Health care professionals and their staff, including hospital staff, are often frustrated by having to complete multiple forms requesting the same or similar information.

Minimizing liability. Health care professionals may be concerned about their liability if a registry exists. What is their responsibility and liability for checking a database to determine if a child needs a particular service? What is their responsibility and liability for

updating a registry after a patient encounter? How timely should those updates be?

Compatibility with existing systems. Many health care professionals have installed computerized office management software. Incompatibility between office and registry software could lead to extra time and costs.

System perspective. The "system" refers to the agencies or organizations that have the following responsibilities: the health of populations living within certain geographic boundaries, the health of those who receive care at a particular facility, and/or the health of those who are enrolled or covered by various insurance programs.¹³⁸ For agencies or organizations with these broad responsibilities, the ideal would include:

Capacity to monitor system performance. This includes the ability to monitor screening coverage, follow-up rates, and health outcomes affected by screening and care programs. It would also include the capacity to provide feedback to individuals or facilities responsible for managing or providing services at each stage in the screening and care cascade. This would enable identification of strengths and weaknesses in the programs to improve overall system performance.

Promotion of collaboration across agencies and organizations. Screening programs encompass a mix of public and private providers; from hospitals where screening tests are performed, to laboratories, to clinics that provide follow-up services. The information system that accompanies a screening program should foster communication and collaboration across the agencies and organizations from family to follow-up program.

Public health monitoring. Public health agencies have a responsibility to track trends in the occurrence and pattern of diseases in the populations they serve. The information system should allow monitoring of the prevalence of disease and the definition of the impact of the screening program on morbidity. It should also allow identification of disease in children not identified by screening (ie, "missed" cases), as well as trends in false-positive results.

Optimal use of resources. Multiple entry of the same or similar information into data systems for different programs represents a duplication of effort and thus extra cost.

Health services research. Monitoring the performance of the overall screening program may yield generalizable information that can be used not only locally but also by others to improve programs.

Data access and confidentiality. Public health agencies have a legal mandate to collect information about programs that they support and diseases that are under their jurisdiction. This requires appropriate access to health information. It may or may not require access to information with personal identifiers. When personal identifiers are stored with health information, it is essential that security measures and confidentiality policies, which protect against unauthorized access and violations of privacy, be in place.

There are a variety of challenges to improve the integration of data systems that support different programs. These challenges include state variations, program-specific systems, costs, independence of heelstick screening programs, and public concerns about government data systems. A number of these issues are being addressed as states implement immunization registries. Topics being addressed in developing these registries include development of policies that define politically permissible levels of integration with other programs as well as responsibilities and liabilities for using and updating registries by health professionals and others.

The current system of categorical programs for newborn health, including independent information management systems, may serve the objectives of individual programs. However, on a broader level, it is inefficient, requires collection of duplicative information, and leads to fragmented services. As a result, there are increasing calls for integration of programs and information management systems. There is an opportunity to take advantage of new information management technologies to improve coordination among the various components of the newborn screening system, as well as improve integration between newborn screening and other related programs.^{11,129,139,140} Efforts to improve the internal or cross-system integration of newborn heelstick screening programs, should be done with careful consideration of program objectives and responsibilities at each level of the cascade of activities, from initial screening to long-term follow-up to system evaluation.^{7,9,18}

Response of the Task Force

The Task Force supports efforts to improve the integration and coordination of public programs that serve infants. The current approach to newborn programs has inherent costs arising from duplication of information collection and fragmentation of activities. Efforts to make programs more cohesive have associated costs as well. Given these costs, initial efforts toward improving integration and coordination should focus on a core group of activities and build, to the extent possible, on existing and successful state models. Although states may be the location for pilot efforts, national leadership and support can assist in development of new models for program integration. Two strategies are sound first steps toward improved coordination and integration:

Assess status of state newborn screening systems

Information is needed on the status of state newborn screening systems. Within heelstick screening programs, information is needed on the capacity to manage and integrate information at each stage of the system. More broadly, information is needed on the relationships among newborn programs, particularly the relationship between screening programs and immunization registries. Substantial effort toward development of information systems is being made in a number of states, including activities funded by the CDC through the development of

immunization registries. In October 1998, the directors of the CDC, HRSA, and HCFA sent a letter to state health and Medicaid directors in support of states' sharing of information across programs, and states' use of categorical funds to enable infrastructure development. To support the improvement of newborn screening systems, it would be useful to know how, whether, and to what extent these programs are involved in activities that are supporting infrastructure development and information-sharing.

Support program integration models

Grants from the HRSA could facilitate and foster the involvement of newborn screening systems in infrastructure development activities in states. Flexible grants would permit states to take advantage of individual strengths and assets. Such grants should encourage states to consider integration of heelstick screening programs with a core set of other newborn programs, including birth registration, immunization, newborn hearing screening, and possibly the WIC program. Because these various activities are supported by different federal agencies, it would be important for the HRSA to collaborate with these other federal agencies such as the CDC and HCFA in developing the grant program.

Task Force Recommendations for Public Health Infrastructure Development

National leadership and federal support are critical to strengthening the public health infrastructure. Flexible funding to support experimentation with activities such as program integration is needed. States with the best practices may lead the way, but a national process to share and promote such practices can facilitate these innovative efforts.

- Federal agencies must take action to strengthen the public health infrastructure for newborn screening.
 - The federal government—acting through the HRSA, CDC, HCFA, AHRQ, NIH, and other agencies—should collaborate to provide ongoing leadership and support for development of newborn screening standards, guidelines, and policies.
 - As the federal unit with most responsibility for newborn screening system development, the HRSA should engage in a national process involving government, professionals, and consumers to advance the recommendations of this Task Force and assist in the development and implementation of nationally-recognized newborn screening system standards and policies.
 - Federal resources should be identified to sustain a NSQAP to assist state public health laboratories. Such assistance must be both sustained and expanded as states adopt new screening technologies and modalities.
 - The HRSA's MCHB should strengthen current mechanisms to improve coordination of infant health programs and initiatives within the state and/or between states, including continuation of

funding in support of newborn screening program reviews.

- State public health agencies should direct their newborn screening program to be consistent with professional guidelines and recommendations. Each state public health agency should take responsibility for systems development. Specifically, states and their agencies have responsibility to:
 - Design and coordinate the newborn screening system;
 - Adhere to nationally recognized recommendations and standards for the validity and utility of tests. State newborn screening systems have a responsibility to review the appropriateness of existing tests, tests for additional conditions, and new screening technology and modalities; and
 - Adopt standards for laboratories, health professionals, and health care financing plans based on nationally recognized standards and guidelines for follow-up, diagnosis, and treatment.
- State public health agencies, working under legislative authority, have the ongoing responsibility to ensure quality and evaluate program effort. States and their state public health agencies should:
 - Maintain a newborn screening system that has appropriate evaluation, performance monitoring, and quality assurance activities from initial screening, through follow-up, diagnosis, treatment, and services through adolescence and adulthood;
 - Conduct oversight of program operations, including those outside the public health agency, such as test analysis and tracking, private sector

collection and transmission of screening data, laboratory quality, and the quality of the diagnostic procedures and treatment programs at pediatric subspecialty clinics; and

- Monitor and evaluate program performance through collection, assembly, analysis, and reporting of data, including outcome evaluations.
- States and state public health agencies should implement mechanisms to inform and involve health professionals and the public. Each state should:
 - Develop a program advisory board that is multidisciplinary, involves pediatricians and other primary care health professionals who provide medical homes for children, pediatric subspecialists, and has meaningful representation of families and the general public; and
 - Design and implement public, professional, and parent education efforts regarding newborn screening.
- States and state public health agencies should provide support for coordination and integration of program activities, including information and services. This will require public-private, federal-state, and intrastate partnerships. States should:
 - Use public and private resources to fund demonstration programs that can serve as a testing ground for linking information and services in ways that improve the newborn screening system; and
 - Structure interagency coordination to maximize resources and to improve the efficiency and effectiveness of newborn screening systems.

III. PROFESSIONAL AND PUBLIC INVOLVEMENT

The smooth functioning of a newborn screening system requires the concerted and dedicated effort of its multiple stakeholders. These key stakeholders include health professionals, parents, and the public.

The Role of Health Professionals in the Newborn Screening Process

Newborn screening is one among a group of public health activities conducted in close cooperation with health professionals. Although public health and private medicine have a long history of “unstable coexistence,” stronger linkages have been proposed and are increasingly valuable in the current health care system.^{65,95} Moreover, newborn screening and other public health programs targeted toward the care of infants, face the challenge of assigning responsibilities to a pediatrician or other primary care health professional who may not be identified in hospital records, or may not have been selected by parents at the time of birth. Even if that individual can be identified, he/she may not be well-informed about newborn screening, genetic conditions, infant hearing screening, and so forth.

Those who provide medical homes for children must understand the newborn screening system, apply appropriate professional standards to their practice, and assume responsibility for their role in that newborn screening system.^{46,47,64,141–144} Ideally, the pediatrician or other primary care health professional who is the source of a child’s medical home should take responsibility for the coordination of the newborn screening process, from initial screening through diagnosis and treatment. Thus, involving these health professionals in newborn screening, including test decisions, follow-up, diagnosis, treatment, and evaluation, is of vital importance to the success of the system.^{64,144}

Roles Related to the Testing Component

Ensuring that all newborns receive appropriate screening tests is central to the effectiveness of any newborn screening program. The awareness, knowledge, and practices of health care professionals who provide obstetric and pediatric care are critical to appropriate screening. For infants born in the hospital, a blood specimen or other test information should be obtained from every neonate before discharge or transfer from the newborn nursery, regardless of the nature or status of the infant’s feeding or age, and transmitted to the state screening system. Moreover, for those discharged early (before 24 hours), a repeat blood specimen for some metabolic screening is recommended in professional and public health guidelines. Preterm infants, those being treated for illness, and those born outside a hospital should have newborn screening tests done before the seventh day of life, and before any blood transfusion.^{9,46,47,142}

Roles Related to Follow-up

The rapid follow-up of the infant with a positive initial screening test is the highest priority. The fol-

low-up process requires timely analysis of test results, rapid communication with the state public health agency’s follow-up staff, and communication to the hospital of birth, the infant’s pediatrician/primary care health professional, and/or the pediatric subspecialist responsible for subspecialty follow-up and management.^{6,7,9} State legislation and regulations vary, but most programs require that a health care professional be notified of the test result. This may include the infant’s medical home, the submitter of the specimen, the birthing facility, the physician of record, and/or the subspecialist responsible for follow-up. Programs should require notification of the parent or guardian as well.

When they are notified, pediatricians, family physicians, nurse practitioners, or others play a critical role in this process. They have a responsibility to ensure that any infant with a positive or equivocal screen result is located, retested, and has a diagnosis confirmed or excluded. Unfortunately, because of the rarity of most conditions screened with heelstick blood samples, many health professionals may not be aware of all aspects of the newborn screening protocols in their states. Although virtually all pediatricians indicate they receive positive screening results in a timely fashion, most do not receive the newborn screening results for all infants in their care.⁴⁷ In addition, the majority do not follow up to secure missing results from newborn screening, assuming that the screening test is negative. In these cases, there is no documentation of newborn screening test results.

When a screening test is positive and a diagnosis is confirmed, the primary care health professional has the responsibility to connect the child to the treatment and care management components of the newborn screening system. This is crucial to ensure optimal outcomes and to avoid preventable consequences of the disorder. The most effective methods of locating and following infants with positive initial screening results will depend on local conditions and resources. Public health staff, including public health or community-based nurses, may play an active role in finding, informing, or linking families. Whatever the method, information that identifies a primary care health professional to a specimen can help avoid delays in the follow-up process. The time frame for following infants will also vary by the type of disorder, and by the magnitude and probable significance of the screening test abnormality. Timely follow-up is important for all disorders but is especially urgent for maple syrup urine disease, galactosemia, and congenital adrenal hyperplasia; these disorders can be fatal if not treated soon after birth. While all positive initial screening results must be followed to resolution, every attempt should be made to minimize the anxiety of the family and the emotional and fiscal costs of the inevitable false-positive tests. The primary care health professional can provide counseling and anticipatory guidance to families as they go through the newborn screening follow-up process.^{9,46,47,69,144,145}

Roles Related to Diagnosis Confirmation

Confirmation of presumptive positive newborn screening test results is always necessary. This requires qualified clinical and laboratory assessment of the infant by a pediatric subspecialist and laboratory in a time frame appropriate for the disorder. All diagnostic test results, normal and abnormal, must be reported to the follow-up and evaluation components of the newborn screening system including: the pediatrician or other primary care health professional, the parents, the state follow-up program, and the state laboratory. Many conditions identified by newborn screening programs are complicated by clinical heterogeneity, and thus, specialized diagnostic interpretation and individualized treatment are required. All inadequate or equivocal test results must be considered for follow-up, until determined to be negative by repeat testing or diagnostic evaluation.

Roles Related to Securing a Medical Home

Every child should have a medical home where care is accessible, family-centered, continuous, comprehensive, coordinated, compassionate, and culturally competent.^{64,144} Prospective parents can benefit by identifying a medical home for their child by the end of the sixth month of pregnancy, thus facilitating access to necessary care for the newborn. When the medical home is identified on birth records, follow-up for newborn screening tests is simplified. For most children, the ideal medical home is a pediatric health care professional working in partnership with the child's parents. For children with diagnosed genetic conditions, the source of the medical home should be the most medically appropriate pediatric specialist or multidisciplinary team, working in partnership with a primary care practice. Each patient and family is entitled to the medical home that best addresses his or her specific health care, as well as primary and preventive, needs.

Role of the Medical Home Health Care Professional

The primary care health professional should:

- Review and be aware of the policies and procedures of their hospital regarding all components of screening including the collection and handling of specimens, recording of identifying information, and timely transportation of specimens to the newborn screening program;
- Establish an office protocol to retrieve results of newborn screening for all newborns admitted to the practice when scheduling the first appointment. If screening cannot be documented, then these infants should be screened;
- Follow positive screening results to diagnosis (ie, confirmed or excluded) and report back to the newborn screening system, including repeated screening and diagnostic test results;
- Recommend and ensure access to subspecialty care and care for other illnesses, understanding that this may need to be provided by pediatric health care professionals and facilities with appropriate

expertise for the child's condition and special needs, and may require additional financing;

- Assist the family in understanding the diagnosis, symptoms, and potential implications of the condition, as well as the availability of genetic counseling, family testing, and other family support services. Reassurance should be given to families when an equivocal or positive result proves to be false. Culturally and linguistically appropriate educational materials should be used;
- Coordinate a seamless integration/communication/partnership with the pediatric center of expertise and community services;
- Understand their clear and defined role in providing the medical home;
- Provide health care supervision and preventive care including immunizations, growth and developmental assessments, and patient and parental counseling about health and psychosocial issues;
- Maintain a central record and database containing all pertinent medical information about the child. This record should be accessible to the family and those involved in the child's care, but confidentiality must be ensured; and
- Participate in training and continuing education offered by state programs, and report information such as health outcome data to state newborn screening programs.

Roles of the Subspecialist/Subspecialty Center

Subspecialty health care professionals should:

- Be experienced and knowledgeable about newborn screening and the diagnosis of the conditions targeted by the newborn screening program;
- Be experienced in the long-term management of infants affected by chronic conditions;
- Designate subspecialty care teams that offer appropriate personnel and services, depending on an infant's condition. Examples include: medical expertise; other health care professionals, such as advanced practice nurses, genetic counselors, social workers, metabolic nutritionists, etc; service coordination/case management; and family support services including peer support and other services such as financial assessment and counseling;
- Formulate short- and long-term therapeutic goals, systematic data collection, and outcome evaluation with linkages to the state newborn follow-up program;
- Provide appropriate follow-up information to pediatricians and other primary care health professionals, families, and the newborn screening system; and
- Assume the role of the medical home, with families and in partnership with the primary care health professional, if appropriate.

Roles of the Public in the Newborn Screening Process

State oversight of newborn screening and other public health programs may be structured in a variety of ways. Legislative oversight to monitor compliance with state law is one level.¹⁴⁶ In carrying out

their oversight responsibilities, state officials should use mechanisms to involve consumers. At the state level, a combination of approaches may provide the most effective participation. Mechanisms for addressing specific questions or decisions might include public meetings, workshops, or focus groups.¹⁴⁷ A state commission or similar entity is valuable to conduct ongoing oversight of the newborn screening system.

Public involvement in newborn screening systems has been widely recommended. *The Task Force on Genetic Testing* recommended that: "Consumers should be involved in policy (but not necessarily in technical) decisions regarding the adoption, introduction, and use of new, predictive genetic tests."²⁹ The CORN calls for at least one advisory committee that includes consumer representation in each state.^{7,9} A survey of state newborn screening systems, however, found that only 26 of the 51 jurisdictions reported having consumer representation on advisory committees.⁶ The roles and rights of parents in these public health agency programs varied markedly in terms of the type of information and the consent policies.

The NAS recommended in 1975 that public agencies use commissions to guide state decision-making.²⁰

The 1994 IOM report also recommended having a body independent of the state program or newborn screening laboratory. Such an advisory body would be involved in making decisions about new tests and testing technologies, program evaluation, quality control, and consumer protection activities. Membership of such an entity should include health professionals, experts, families affected by screening, and members of the broader public.²⁸

Roles of the Family

The Task Force recognized the importance of family involvement in newborn screening systems. Parents were involved in developing the recommendations of this report through membership on the Task Force, work groups, and by providing public comment. Based on the input received from parents and consumer advocates, the Task Force concluded that:

- Families should be educated about newborn screening. Information should be provided before birth or after birth. Information should be provided during the follow-up process, if the initial screening test is positive.^{23,148,149}
- Out of respect for the importance they play in the life of a child, the family should be recognized as an integral partner in the health care system. The family is responsible for adherence to recommended interventions and for maintaining contact with their primary care health professionals and pediatric subspecialists.
- The family should be involved in informed decision-making beginning with the initiation of newborn screening through the steps of the positive test result from the initial screening test, the confirmatory testing, and the enrollment in therapeutic interventions.

- Patient educational materials should be developed and reviewed in conjunction with families, be assessed for literacy levels, and reflect cultural competency.
- Families should receive information and counseling so that they are aware of the diagnosed condition, the potential associated co-morbidities, the short- and long-range treatment goals and interventions, and the availability of health care resources, including primary care health professionals, pediatric subspecialty consultants, genetic counselors, and state financial case management and assistance programs.
- Affected individuals and families should be involved in newborn screening program oversight (eg, advisory boards, review committees).

Professional and Public Involvement in Informed Consent

The Debate Over Informing and Consent

The issue of educating and informing parents about, and receiving permission for newborn screening is not simple. This issue has been debated since the first mandatory metabolic screening program for PKU began in 1963. In 1994, the IOM report raised concerns about the addition of unproven tests and made a recommendation for using informed consent when newborn screening tests or testing methods have not been studied carefully.²⁸

During the past 5 years, these recommendations have been discussed and debated by public health professionals, parent organizations, ethicists, and others. The IOM Committee's recommendations were introduced as "somewhat ideal scenarios" (preface) and it was recognized that such practices might not be realistic. Moreover, the Committee did not reject the idea of mandatory screening for conditions such as PKU or congenital hypothyroidism where tests and treatment have been proven safe and effective.

In response to the IOM recommendations, the Newborn Screening Committee of the CORN, the American Society of Human Genetics, and the Joint Committee on Professional Practices of the American College of Medical Genetics raised further questions about the practicality of implementing informed consent policies.^{150–153}

Current State Practices

State policies regarding informing parents and parental refusal and consent vary widely. Forty-nine states have specific legislation that requires newborn screening; 3 states have provisions for informed decision-making.^{23,146,154} Currently, Maryland has a voluntary newborn screening program, Wyoming uses an informed consent model, and Massachusetts recently began using an informed consent process in a pilot newborn screening program. Most states permit parental refusal, but only under limited circumstances. Parents may not be told directly that they have the opportunity to refuse, and for some parents, mandatory offering may be confused with mandatory screening. In most states, it is routine practice to

accept parent refusals but not to ask for documentation (ie, with a form and parental signature).¹⁵⁵

There are several arguments in favor of not seeking parental permission for newborns screening.¹⁵⁶ First, and perhaps most important, is that screening and potential detection is in the interest of the child and the parents' objections should not hinder that screening process. This may be more compelling for PKU than for diseases where the benefits of screening would be less clear-cut, as with Fragile X syndrome.^{157,158} As most state newborn screening laws make accommodations for parents who refuse testing, this argument does not seem to be the basis of the current approach. A second argument is that it is not feasible or it is too costly to talk to parents and ask permission. In early studies of the Maryland newborn screening system, the cost and time involved in the Maryland program did not appear to be prohibitive. The current approach in Maryland is a simple "goodwill" informed consent for the total screening package and does not allow for separate consent or refusal for each disorder.¹⁵⁹

Shared Decision-Making as a Model for Informed Consent

An informed consent process for medical procedures and interventions is a basic expectation of the general public today. Although it is often equated with signing an "informed consent form," shared decision-making can occur without signing a form, and signing a form does not guarantee that shared decision-making or informed consent has occurred. Shared decision-making refers to a conversation, between the health professional and the patient/parent, where relevant information is disclosed. Most of the discussion between professionals and parents regarding the care of children is rather informal.^{107,155,156,160} Nonetheless, health professionals talk with parents not only because they have to, to treat the child, and not just because they may think that the parents will be more "compliant" if they buy into the plan; but more importantly because health professionals respect the independent and important role parents play. For this reason the Task Force emphasized the importance of the conversation, not the documentation to achieve shared decision-making.

The Task Force recommended that additional approaches to informing and educating parents be studied further. A greater emphasis on parental education may improve parent understanding and increase the number of parents who comply with recommendations for further testing and follow-up. Such education may also help parents deal with the anxiety associated with equivocal results, repeated tests, and false-positive results. Furthermore, informed decision-making is particularly important when the safety and effectiveness of some newborn screening tests and screening technology are still being evaluated. With the addition of new DNA-based tests, and the addition of screening tests for conditions for which the treatment intervention or the efficacy of the treatment intervention is unknown, the ethical, legal, and social demands to ob-

tain documentation of permission for newborn screening may increase.

The consensus of the Task Force is that the goals of newborn screening can be accomplished while acknowledging the role parents play in deciding what is going to be done to their children, and while also respecting the wishes of those few parents who object. The Task Force achieved a new level of consensus about consumer information, along with recommendations for future action. Parents need to be informed about the benefits and potential risks of the tests and treatments, the policy for storage and use of specimens, and the mechanism by which families will receive test results. Of particular importance in informing parents is their understanding of why they should respond to abnormal results, how to respond, and the possibility of false-positive results. Determining the best mechanisms to inform parents and promote screening then becomes the issue. All prospective parents should be made aware of the newborn screening process. One practical strategy for educating parents is for prenatal health care professionals to provide this information early on during the course of prenatal care. Ideally, this could be accompanied by educational material and/or videotapes provided during one of the third trimester prenatal visits, with a brief review by office or clinic staff.

Task Force Recommendations to Increase Professional and Public Involvement in Newborn Screening Systems

The *Task Force* recommends that:

- The pediatrician or other primary care health professional who, in partnership with parents, is the source of the child's medical home, should:
 - Ensure that all newborns admitted to their practice have received adequate newborn screening, and that appropriate documentation of this testing is present;
 - Follow positive screening results to diagnosis (ie, confirmed or excluded), including repeated screening and diagnostic testing;
 - Coordinate a seamless system of care with pediatric subspecialty clinics, tertiary care centers, and/or community-based providers, when a child is diagnosed with a disorder through newborn screening;
 - Maintain a central record and database containing all pertinent medical information about the child. This record should be accessible to the family and others involved in the child's care, but confidentiality must be assured; and
 - Assist the family in understanding the diagnosis, symptoms, and potential implications of a diagnosed genetic/metabolic condition, as well as the availability of genetic counseling, family testing, and other family support services.
- Parents should receive information (on behalf of their children) about newborn screening.
 - Prospective parents should receive information about newborn screening during the prenatal period. Pregnant women should be made aware of

the process and benefits of newborn screening and their right of refusal before testing, preferably during a routine third trimester prenatal care visit.

–Parent knowledge should be reinforced after delivery by educational materials and discussion as needed by the infant’s primary care health professional and/or knowledgeable hospital staff.

- Prenatal health care professionals as well as the infant’s primary care health professional should be knowledgeable about their state’s newborn screening program through educational efforts coordinated by the state’s newborn screening program in conjunction with a newborn screening advisory body.

- Written documentation of consent is not required for the majority of newborn screening tests, for example, those tests of proven validity and utility.

–Parents should always be informed of testing and have the opportunity to refuse testing.

–If after discussions about newborn screening with health professionals, parents refuse to have their newborn tested, this refusal should be documented in writing and honored.

–If a newborn screening test is investigational or in the process of being developed, the benefits or potential risks have yet to be demonstrated, and identifiers are not removed from the specimen, informed consent should be obtained from parents and documented.

- Studies should be done to broaden understanding of the ways in which communication can be done more effectively for the benefit of consumers.

–Pilot studies and evaluation research should be conducted to assess the potential impact of revised parental permission and informed decision-making policies.

–Each state or region should, with input from families who have children with special needs and/or parent information centers, develop and provide family educational materials about newborn screening.

–Evaluation of materials should be ongoing, par-

ticularly because of the changing demographics of childbearing, cultural changes, and rapid developments in genetic science.

- Parents have a right to confidentiality and privacy protections for the medical and genetic information in any type of newborn screening results. Based on nationally recognized standards and guidelines, each state should have appropriate policies and mechanisms in place to ensure families’ privacy and confidentiality. Laws to guarantee genetic privacy and protect against genetic discrimination should benefit patients identified by newborn screening.

- States and the federal government should include public participation in medical policy-making. The SACGT provides a mechanism for public participation in genetic policy development at the federal level. Each state should establish and fund a newborn screening advisory body with public participation to advise on newborn screening policy developments.

–Such an entity should include a broad range of public advisors representing parents, health professionals, third-party payers, appropriate government agencies, and other concerned citizens.

–Such an entity should be empowered to advise state officials about screening for particular conditions based on accepted standards and be consulted about the development of related state regulations.

–Such an entity should be involved in the review of new tests under consideration by the state and in the development of pilot programs for new tests.

–Such an entity should be involved in the ongoing evaluation of all aspects of the state’s process for newborn screening. Oversight activities should include a review of: testing, follow-up and treatment efforts; the impact on families of receiving a false-positive screening result; and the state’s process for handling consumer input including grievances.

IV. SURVEILLANCE AND RESEARCH

Public health agencies must ensure adequate policies for surveillance and research related to newborn screening. Surveillance and research are important activities that impact the growth of the newborn screening system. Without surveillance activities, such as performance measurement or outcome evaluations, it is difficult to assess the degree to which a particular newborn screening program benefits infants. Without research to determine the effectiveness of new technology or to develop new screening tests, potential benefits to newborn screening systems may be lost.

Performance Measurement

State and federal public health agencies engage in both the collection and analysis of data in their assessment activities, and for a variety of reporting requirements. In particular, state MCH programs should be involved in the design, implementation, coordination, and evaluation of newborn screening systems, as they are the locus of responsibility for child health. Each state MCH program has a variety of care and services aimed at the population of CSHCN. Because children with conditions identified through newborn screening are a subset of CSHCN, achievement of the "National Agenda for Children With Special Health Care Needs" will improve newborn screening systems and services. The core objectives for outcomes of this National Agenda are:

- All CSHCN will receive regular ongoing comprehensive care within a medical home.
- All families of CSHCN will have adequate private and/or public insurance to pay for the services they need.
- All children will be screened early and continuously for special health care needs.
- Services for CSHCN and their families will be organized in ways that families can use them easily.
- Families of CSHCN will participate in decision-making at all levels and will be satisfied with the services they receive.
- All youth with special health care needs will receive the services necessary to make appropriate transitions to all aspects of adult life, including adult health care, work, and independence.¹⁶¹

The HRSA's MCHB has a key role to play in assisting states to work toward these objectives. The MCHB assists in measuring performance and stimulating the development of newborn screening systems and related information systems, with a focus on development of standardized data sets, outcome evaluation, and analyses of cost-efficiency and effectiveness. Federal guidance issued in 1998 established the HRSA's Title V Block Grant Measurement Performance System, and required that state MCH agencies report on a set of 18 national core performance measures. Among these core performance measures are measures directly related to newborn screening and the recommendations presented in this report.

These performance measures require each state to assess the extent to which they:

- Increase the percent of newborns in the state with at least 1 screening for each of PKU, hypothyroidism, galactosemia, hemoglobinopathies (eg, sickle cell disease) (combined).
- Increase the percent of newborns who have been screened for hearing impairment before hospital discharge.
- Increase the percent of CSHCN in the state who have a medical/health home.
- Increase the degree to which the state ensures family participation in program and policy activities in the state CSHCN program.¹⁶²

States also have defined additional performance measures that fit with their priorities, programs, and populations. State-defined indicators are selected to measure the percent of newborns who receive additional newborn screening tests, the rate of selected congenital conditions (for birth defects surveillance), and the percent of identified infants who have received follow-up care and treatment within the medical home.¹⁶³

The strategic goals and objectives of the MCHB are linked to these performance measures. In relation to newborn screening, the MCHB objectives aim to do the following by 2003: ensure that all newborns are screened, diagnosed, and provided treatment for disorders identified by state specific newborn screening programs; ensure that 50% of all children, including CSHCN, are enrolled in a medical home; and ensure that 100% of the major national managed care organizations have a mechanism to measure the quality of the components of a medical home for CSHCN. Other objectives aim to enhance research and surveillance capacity such as increased use of data and information, improved scientific knowledge base, and use of linked electronic databases.

Performance measures also have been developed for the private sector. For health plans, and the professionals and facilities that deliver care for their enrollees, the Health Plan Employer Data and Information Set (HEDIS) sets out performance measures.¹⁶⁴ The HEDIS is currently being used by a wide range of private and public (ie, Medicaid) purchasers who seek to measure the value and performance they receive for the dollars they spend on coverage. In terms of newborns, the HEDIS includes measures on immunization rates, low-birth weight rates, and length of newborn stay; however, it does not assess performance on newborn screening. Other measures are being developed in the private sector, including a set specifically for CSHCN.

In a health care system that demands increasing accountability from government, health plans, and health professionals, these goals, objectives, and performance measures help to define surveillance and research needs in newborn screening systems.

Program Evaluation

Setting Priorities for Data Collection

The integration of information systems would allow newborn screening program evaluation to take

place with more ease. Ideally, the information obtained by a newborn screening program would allow the description of:

- The number and percent of children
 - adequately screened,
 - with appropriate follow-up,
 - with false-positive and false-negative results,
 - with specific diagnoses, and
 - with appropriate care.
- The time between the newborn screen and the initiation of treatment.
- The long-term improvement in health status occurring as a result of screening, follow-up, diagnosis, and treatment.
- The number of children diagnosed with a condition missed by the screening programs and, where possible, an assessment of the reasons they were missed.
- The number and percentage of children lost to follow-up.

Improving Information Systems for the Purposes of Surveillance and Research

Data collection and analysis are necessary for surveillance activities, epidemiologic studies, program evaluation, and research. These activities require a strong commitment to developing and maintaining an adequate information infrastructure.

In considering the improvement of data systems for newborn screening programs and coordination with other programs, an initial state assessment, consisting of the following components, is necessary:

- The objectives of the program.
- The components of the system and responsibilities for managing each component.
- The collection and intended flow of information.
- The quality of communication among persons and facilities at different stages of the program.
- Procedures for follow-up (short- and long-term).
- Information required for evaluation of the program.

These components of newborn screening systems are outlined more fully in the CORN system guidelines.^{7,9} For other types of newborn screening, such as hearing screening, the strength and integration of information systems are equally important. Descriptions of these components in the context of an individual state program will enable a clearer articulation of the purposes of the information system needed to support these elements of the program.

Some broad concerns underlie the development of information systems, and these concerns assume greater importance as complex systems for record linkages and system integration are proposed. These include:

- Ensuring those information system procedures relate to program objectives.
- Defining time frames for data collection and feedback (eg, how quickly should data be accessible at each stage in the screening, follow-up, diagnosis, and treatment process).

- Defining reporting procedures (eg, what reports will be made, who will receive them).
- Ensuring commitment to maintaining systems.
- Ensuring that procedures for maintaining, transmitting, analyzing, and disseminating data conform to ethical guidelines and legal standards.

The Role of Record Linkages

Record linkages, or the process of relating information about individual newborns from different information systems, provides an approach to integrating information management within a program (eg, information from various stages in the service cascade) and integrating information across programs. In some instances, such linkages may not be required to meet specific objectives; instead, a capacity to synthesize information from multiple sources would be sufficient.

Issues to consider for record linkages or information synthesis include:

- **Short- and long-term information needs of the screening program (eg, screening and follow-up data).** This would include information needed to optimally serve families, to assess the newborn screening program, and to provide information that would improve the operation of screening and follow-up.
 - Definition of screening coverage requires a denominator, which is defined by the total number of live births. This would involve relating newborn screening data to the birth certificate file.
 - Assessment of health outcomes involves follow-up of infants with diagnosed disorders through use of medical records. This may require use of multiple data sources from health care professionals (eg, hospital discharge records, outpatient visits). Newborn screening systems alone often lack the authority and personnel to collect outcome data.
- **Integration with other data systems to minimize duplication and facilitate cross-program communication.** This would require definition of core data sets that could be better coordinated or integrated, (eg, electronic birth files and immunization registries). One example of a core data set is birth registration, newborn heelstick screening, newborn hearing screening, and immunization; these are activities that are initiated in the newborn hospital nursery and are universal (or likely to become universal). Information from the WIC program, which serves a large proportion of infants, and birth defect registries may be other data sources to consider as part of the core.
- **Definition of the purpose of record linkages and data synthesis.** The purpose and intended uses of a data system will have a profound impact on its level of technical complexity and cost, depending on whether the intent is to:
 - Allow retrospective program assessment using historical data, such as an annual assessment, or
 - Improve screening and care management through real-time data systems.

In proposing development of an integrated information system, there are multiple technical and logistic considerations that should be taken into account. These procedures for accessing and manipulating screening files include:

- **Distillation of records on samples to individual client-level data** (ie, ensuring that multiple screening or follow-up samples can be identified as belonging to an individual child).
- **Definition of variables and criteria that would be used to define linkages.** Criteria for associating records from heelstick screening to the birth file must be defined, including acceptable levels of unmatched (sensitivity) or mismatched (specificity) records. The role and definition of a universal health identification number is beyond the scope of this report but is critical to any discussion of the integration of health systems. Clearly, the use of a standard identification number would greatly facilitate the integration of data systems across newborn service programs. States have developed numbering systems that serve newborn screening programs and have piloted systems that would allow use of this number as a prototype for a broader, health identification number. In addition, substantial work is being done nationally in consideration of a standard health identification number. The implementation of such an identification number would not solve all problems in merging information across programs, because errors may be made in its entry into records or databases.
- **Consideration of the role of new technologies for identification and information storage.** In some hospitals “bar code” technology (eg, on wristbands and forms) is being used to facilitate and ensure identification of newborns. The use of scanning devices at the time that various procedures are performed to collect samples (heelstick), perform other tests (hearing screen), or provide service (immunization) offers one approach to integrating information management at the hospital level.

Another technology is the use of so-called “smart” cards, credit card-sized information storage devices that allow the creation of a highly portable record that would be carried by parents and updated or read by professionals at various points of service. This technology could support a highly decentralized information storage and retrieval system that, in itself, would support some of the above objectives (eg, improved health professional access to patient information) but not all (eg, public health monitoring) in the absence of linkage with more centralized systems.

Using an Evidence-Based Approach to Make Decisions About New Tests

Since the 1960s, decisions about which tests to use in newborn screening programs often have been made in an extemporaneous fashion, depending on recommendations from professional groups, patient advocates, state legislators, and newborn screening

programs.^{19,35,77,84,157,165–172} Only rarely, for example with screening for sickle cell disease, has the decision been based on empirical evidence of safety and efficacy from a clinical trial (and in that case, the clinical trial findings were related to the effectiveness of treatment).^{16,29,41} Surveillance and research are essential to provide the evidence needed for state-level decisions and nationally recognized standards.

The Task Force on Genetic Testing in their report, *Promoting Safe and Effective Genetic Testing in the United States*, gave particular attention to an evidence-based approach.²⁹ They recommended that a test must be determined to have analytical sensitivity and specificity before it is made available in practice. Clinical validation is the next step, with clinical sensitivity, specificity, and predictive value determined through study with a sample population that is representative of the test’s target population. The test should also have clinical utility—that is, interventions to improve the outcome for the infant must be safe and effective.

In making decisions about which newborn screening tests to use and for whom, states need information. Pilot studies are an important tool in this process.^{59,173} Such studies might be undertaken by an individual state (eg, currently several pilot studies are underway in Massachusetts). For rare conditions, collaborative efforts between states will be needed to expedite data collection. Collaborative clinical trials (such as the prospective study of prophylactic penicillin with sickle cell anemia) may also be needed to evaluate the effectiveness of treatment and interventions. In all such studies, safeguards are needed to protect the confidentiality of the individual infants who are the source of the data.

The Task Force on Genetic Testing called for an active role by federal agencies, particularly the NIH and CDC, in supporting collaborative efforts to collect data on the safety and effectiveness of genetic tests.²⁹ Support might be in the form of funding, guidelines, and/or oversight.

Establishing Policies and Procedures for Use of Residual Blood Samples in Research

In the case of newborn heelstick testing, data collection and analysis activities also require that policies and procedures be in place to cover the use of residual blood samples for research. Such research might be related to new or existing newborn screening technologies, or to epidemiologic research relevant to clinical medicine and public health. In either case, state policies should determine storage conditions, uses, and consumer protections.

Almost all infants screened have residual blood samples retained by the state programs. Enough blood is obtained when performing heelstick newborn screening to permit programs to repeat tests when necessary. However, because repeat tests are not always necessary, and a repeat test may not use up the blood sample, the vast majority of infants screened (in excess of 95%) will have residual blood samples retained by the state programs. Currently, state programs hold these samples for variable lengths of time: 10 programs save samples for 21

years or more; 6 programs for 5 to 7 years; 2 programs for 1 to 3 years; 6 programs for 6 to 12 months; 21 programs for 1 to 6 months; and 5 programs for 1 to 4 weeks. Only 1 program is known to save the samples under optimal conditions for later use in biochemical analyses. Optimal storage conditions are much less critical for genetic analyses, and samples stored in many states are adequate for genetic analyses. The lack of uniformity between programs reflects uncertainty and debate over whether residual blood samples should be retained and, if so, for how long and under what conditions.^{6,174–177}

Potential Uses of Residual Blood Samples

The conclusions of the Task Force are predicated on 2 principles: 1) that residual blood samples are a valuable resource, and 2) that their use should be carefully managed to protect the interests of those from whom they are obtained. To achieve the maximum benefit from residual blood samples, and to ensure that samples are used to promote child, family, and public health, state programs (with input from the public) should thoroughly evaluate their policies and procedures. The retention of these samples should be guided by state policies that articulate the objectives of the storage and use of the samples, and include safeguards against inappropriate use. These policies should define the rationale for retaining and analyzing samples with or without identifying information. However, policies and protocols for the retention and use of residual blood samples should not hinder the primary function of newborn screening programs, which is the early detection and treatment of infants with conditions that the newborn screening program has targeted to screen.

Residual blood samples may be used for several purposes including:

- **Research related to new or existing newborn screening modalities.** As technology advances for newborn screening programs, new testing modalities will be developed for conditions included in current programs, and new tests will be added for other health conditions. Residual heelstick blood samples can be used to ascertain the validity of new testing modalities for existing conditions and of tests for new conditions. Identifiers are sometimes retained to enable follow-up contact with an infant's family if an effective intervention is available for children diagnosed with the condition.
- **Epidemiologic research relevant to clinical medicine and public health.** Public health officials need population-based data to determine the appropriate allocation of resources to care for children with specific conditions. A thorough understanding of many health conditions requires epidemiologic data on the prevalence of specific genetic or biochemical attributes in the general population. Residual newborn screening samples constitute a specimen bank of a large cohort of the population of states. As genetic technology advances, such a comprehensive bank, linked to basic demographic information, may be useful for certain types of research. The potential utility of

such a resource will need to be carefully evaluated because residual blood samples in this context will not be linked to clinical data on the children.^{178–186}

- **Clinical or forensic testing.** For children who have moved and cannot be located, the heelstick blood sample may represent the only source of a biological specimen from a given child. The sample may be useful for forensic purposes. Testing of residual blood samples may be essential in the postmortem identification of a genetic condition that may have contributed to a child's death. At least 1 state has decided to store newborn blood spots indefinitely to permit identification of children who have been kidnapped.¹⁸⁷

Ethical Concerns Related to Use of Residual Blood Samples

Storage and use of residual newborn screening blood samples raise a number of practical and ethical challenges. Ethical challenges include the development of guidance regarding the use of residual blood spots for purposes other than those for which they were originally obtained. The protection of privacy and confidentiality among children and families is a serious concern. In the case of newborn screening, when blood samples are collected from infants as a matter of law, there is additional reason to ensure appropriate storage and use.^{175–177,188–191}

At the same time, residual newborn screening samples have been used to address important public health issues. The prevalence of in utero exposure to drugs and environmental agents; the allele frequency of genes associated with significant morbidity, mortality, or disability in infancy or childhood,^{66,157,192} and the prevalence of serious maternal or intrauterine infections have been determined in various populations by anonymous use of residual blood spots.^{178,179,183,184} Samples linked to outcome have been used to assess the feasibility of screening for various diseases of the newborn and infant, and to determine risk factors for birth defects and developmental disabilities.^{180,185,186,193} To date, there have been no published reports of misuse of residual newborn screening samples in research projects; however, the potential for use and misuse is expanding.

The Task Force recognized the ethical challenges in a new era of genetic science and the practical challenges related to cost, space, storage, and the development of databases to catalog large numbers of samples. The Task Force also discussed the potential value of these samples for research and also recognized that their use for research must include protections for the privacy and confidentiality of children and their families, as would be the case for any research with human biologic materials. There is active debate in the US health care community about the appropriate uses of residual human biologic materials. Policies and procedures for the use of residual newborn screening samples need to be developed in the context of this debate.

Defining Sample Categories

One factor affecting the level of risk associated with using human biological materials for research is whether a particular sample can be linked with an

individual. Commonly agreed on definitions that reflect the degree to which samples can identify an individual are important to building an understanding of how newborn screening blood samples are or are not protected. In its evaluation of residual blood samples, the Task Force defined 2 broad categories for use of residual newborn screening blood samples: *unlinked* and *identifiable* samples. Based on statements by the National Bioethics Advisory Commission (NBAC), the Task Force used the following definitions¹⁹⁴:

- Unlinked (sometimes called anonymous) samples lack identifiers or codes that can link a particular sample to an identified specimen or a particular human being. These samples may have originally been collected without identifiers, or the identifying information (eg, names, registration numbers) may have been removed; making it impossible to link the sample with the patient.
- Identifiable samples are either directly identifiable or coded with a link to identifying personal information.
 - Directly identified materials have identifying information (eg, name or patient number) attached and available to researchers.
 - Coded samples are numbered or labeled in a manner that does not allow a researcher using the specimen to identify the individual from whom the specimen was collected. However, a link between the code marker and personal identification information is retained, permitting patient identification for other reasons (such as family requests). In some circumstances, linkage information between samples and personal identifiers can be retained by a third party to strengthen safeguards for privacy and confidentiality.

An important topic of debate is whether consent for research is needed from the individual from whom biological materials are obtained. Although this question is not totally resolved, major efforts are underway across the country to develop mechanisms to inform patients and obtain their consent.

By contrast, 3 states currently require informed permission from parents for newborn screening itself. None of these states obtains specific permission for use of the samples for research purposes; however, the state of Maryland does inform parents in

an informational brochure that samples may be used for certain types of research and individual results will not be identifiable. The universal lack of permission for using bloodspots for research gives added weight to concerns about privacy, confidentiality, and discrimination.

The text and Table 2 below outline 2 broad categories for the use of residual newborn screening blood samples. Together, these reflect the conclusions of the Task Force about appropriate purposes, applications, and protections.

Use of unlinked samples. Unlinked samples may retain limited demographic information (eg, gender and ethnicity) to provide general descriptive categories in epidemiologic analyses. However, such information should not be sufficient to permit identification of an individual. Current national standards stipulate that epidemiologic research can be conducted without consent, as long as identifiers are removed. Parents should be informed that unlinked samples might be used for quality improvement purposes or for epidemiologic research consistent with the goals of newborn screening programs. Protocols for the use of unlinked samples in hospital and laboratory quality assurance activities need not be submitted for institutional review board (IRB) review. Legislative approval and regulatory guidance for research on unlinked samples should be consistent with the goals of newborn screening programs and public health efforts.

Use of identifiable samples. The Task Force concluded that parental permission should be sought for the use of identifiable samples in research to validate tests for additional diseases, or for epidemiologic research. Identifiable samples from newborns should be used for research only if: 1) IRB approval is obtained for the proposed research, 2) consent is obtained from the child’s parent(s) or guardian for the proposed research, 3) newborn samples represent the optimal source of available tissue for the research, 4) unlinked samples will not suffice, and 5) acceptable samples from consenting adults are not available.

In accordance with current federal regulations regarding research involving children, use of such samples for research, that poses more than minimal risk, should be limited to activities that benefit the

TABLE 2. Classification of Biological Samples

Use	Unlinked	Identifiable
Focus	• Populations	• Populations to individual
Purpose	• Epidemiologic studies • Quality assurance • Test refinements	• Medical and clinical studies for individuals • Clinical validation of tests for additional disease • Legal and forensic work
Consent process	• “Right of refusal” permission routinely used in newborn screening	• Informed consent
Protocol review	• Quality assurance studies do not require IRB	• IRB required unless use is routine laboratory quality assurance • Exception is court ordered uses
Other issues	• No markers remain for possible future uses—individual or population	• Concerns about privacy protections and confidentiality may arise

child or that are of importance to understanding a condition affecting children. If a state foresees the possibility that research using residual specimens will be done at a later date, a mechanism should be in place to inform parents of and obtain permission for that research. An up-front mechanism of informed consent, at the point of the heelstick, is one logical way of initiating the process of informed consent. Any research on identifiable samples that is not covered by the original consent would require recontacting the parents. Proposals to recontact patients with specific results should be justified to and approved by an oversight body before contact is made. Forensic (eg, for identification of a missing or deceased child) or clinical uses of the samples should be with the family's consent or with a legal mandate. If identifiable samples are maintained, policies and procedures need to be developed to define appropriate access for the purpose of forensic testing or other legal purposes (see Table 2).

Task Force Recommendations to Strengthen the Infrastructure for Surveillance and Research

The *Task Force* recommends that:

- State MCH programs should conduct a review of the newborn screening system and its relationship to the HRSA MCH Block Grant Performance Measures and evaluate the quality of data of the newborn screening-related performance measures.
- The federal HCFA should develop HEDIS measures to evaluate the health plans' performance within the newborn screening system.
- A federally-funded newborn screening research agenda should be outlined that aims to: develop better tests (more sensitive, more specific, and less costly); assess the validity and utility of new technologies (eg, tandem mass spectrometry, DNA-based testing, and other evolving technologies); and define appropriate uses of residual biologic samples for a population-based research and surveillance.
- The HRSA's MCHB should provide grants to states to stimulate development of newborn screening information systems that are connected to the medical home, with a focus on newborn screening system process and outcome evaluation, development of standardized datasets, analyses of cost-efficiency and effectiveness, and integration with other public health data systems. Support for technological innovation (ie, new test technologies) should include these measures.
- Pediatricians, pediatric subspecialists, and other health professionals who care for children should contribute to newborn screening data collection to advance knowledge about health outcomes and intervention effectiveness. Professional associations, the HRSA-funded National Newborn Screening and Genetics Resource Center, and state

newborn screening programs should develop strategies to assist health professionals in their efforts to participate in and learn from newborn screening information systems.

- Pilot studies should be undertaken to demonstrate the safety, effectiveness, validity, and clinical utility of tests for additional conditions and new testing modalities. Informed consent of parents is called for in all such pilot studies. These studies might be undertaken by individual states, regional or nationwide groups of states, or through federal grants provided to research institutions across the country.
- Federal and state public health agencies, in partnership with health professionals, families, and representatives of ethnic, minority, and other diverse communities, should:
 - Develop model legislation and/or regulation that articulates policies and procedures regarding utilization of unlinked and identifiable residual samples for research and public health surveillance. This process should include review and consideration of the recent recommendations to the President set forth by the NBAC for research involving human biological materials;
 - Develop model consent forms and informational materials for parental permission for retention and use of newborn screening samples;
 - Develop educational materials for parents that includes information regarding the storage and uses of residual samples;
 - Organize collaborative efforts to develop minimum standards for storage and database technology to facilitate appropriate storage of residual newborn screening blood samples at the state level; and
 - Consider creating a national or multi-state population-based specimen resource for research in which consent is obtained from the individuals from whom the tissue is obtained. Such a resource could be an alternative to retaining newborn screening samples for potential use in research.
- Using national recommendations, each state program should develop and implement policies and procedures for retention of residual newborn screening blood samples that articulate the rationale and objectives for storage, the intended duration of storage, whether storage is with or without identifiers, and guidelines for use of identifiable and unlinked samples. An advisory group for newborn screening programs with broad professional and family/community representation is a valuable resource in developing policies and procedures and in reviewing applications for use of retained samples. The advisory body also could determine priorities for use.

V. THE ECONOMICS OF SCREENING

Public health agencies should ensure adequate financing mechanisms to support a newborn screening program. Because states require universal newborn screening for certain conditions, they have an ethical and fiscal responsibility to ensure that children with identified disorders receive maximum benefit from early diagnosis and intervention. A newborn screening system is comprised of 5 parts: 1) screening, 2) short-term follow-up, 3) diagnosis, 4) treatment and management, and 5) program evaluation and quality assurance. Because financing is needed for each component, newborn screening systems need adequate funding to serve all children. Ensuring adequate finances, through public health spending and other funds, is essential.

Defining Cost Effectiveness

Screening is done to prevent disease and its consequences, with the expectation that expenditures now will reap benefits in the future. In making decisions about newborn screening systems, the value compared with the public cost has long been a consideration.^{33,40,81,100,195–197} In addition to having a reliable test and system that can benefit children, public health officials may be asked to justify the cost of population-based newborn screening.

Many believe that screening, as a tool for prevention, is a way to reduce costs.¹⁹⁸ However, screening may increase, not reduce, the cost of a public program. In addition, it may avert costs that otherwise would have been incurred within the health care system as a whole, or outside the health care system. Economists and health policy analysts use 2 types of calculations—cost benefit and cost-effectiveness—to estimate the potential for savings, potential for averting costs, and potential for achieving benefit in reduced mortality and morbidity.

Cost-benefit calculations attempt to value everything, including health effects, in terms of dollars.¹⁹⁸ The cost-benefit of newborn screening for particular conditions is the cost of screening and treatment minus costs averted in dollars.^{4,14,100,199} Although this makes it easier to perform comparisons, many object to the idea that human lives and health can be represented by dollars. Also, there is disagreement about what monetary value to assign. Reaching agreement on the goal of the intervention (in this case screening) also is important—is the goal to save lives, prevent disability, reduce public medical expenditures, or something else?

Alternatively, cost-effectiveness analyses compare the cost of doing something to the cost of doing nothing, or of doing something else. It is useful in showing which alternative is preferable.¹⁹⁸

The Cost-Effectiveness of Newborn Screening

In 1988, the US Congress Office of Technology Assessment (OTA) published a review on the effectiveness and costs of newborn screening for specific conditions, compared with no screening.¹ This review was conducted using the best information available at that time, and was done using a “basic

approach” to newborn screening. The “basic approach” was common to all states, and was defined as collection and testing of a single blood specimen to identify cases of PKU and congenital hypothyroidism. Using the “basic approach”, the OTA analyses concluded that net health care savings per 100 000 infants screened (in 1986 dollars) was \$3.2 million, and that the net health care savings per case detected and treated was \$93 000.

The OTA also compared the cost-effectiveness of the “basic approach” to 6 expanded newborn screening strategies. These new strategies included variations that would test for additional diseases or conduct more intensive screening for PKU and congenital hypothyroidism. Unlike many previous efforts, these expanded strategies included the cost of specimen collection and follow-up. The OTA found that each of the expanded strategies for screening was more effective in detecting affected infants, and more costly than the “basic approach”. Based on their calculations, the OTA found that:

- detecting additional cases by adding tests to an initial (single) specimen is less costly than collecting and analyzing a second specimen, and
- more cases can be detected with repeat heelstick testing (which required a second blood specimen for additional tests or as follow-up to early discharge), but the cost of collecting additional specimens adds significantly to the overall cost.

The report states: “Each of the 6 expanded strategies would save more babies from deadly or disabling diseases than the basic strategy . . . but the incremental costs of achieving those extra successes are high.”¹ This was true whether additional specimens were collected to detect extra cases of PKU and congenital hypothyroidism, to detect homocystinuria, or as a precaution against missed cases. For example, the OTA found that the cost of detecting 1 extra case using an expanded 1-specimen strategy (testing for additional diseases from 1 sample) was about \$85 000. The OTA concluded that “this amount would buy an entire lifetime for a child with one of these disorders, and is low compared with the cost of many therapies currently considered accepted medical procedure.”¹

Notably, the OTA cost-effectiveness analysis did not include newborn screening for sickle cell anemia, biotinidase deficiency, congenital adrenal hyperplasia, as well as other conditions that were being screened for in some states and through pilot programs. Moreover, some believe that the OTA analysis did not fully take into account the public health or personal care burdens for identified conditions. If these factors were considered, the estimated net costs and savings would be different. For example, if additional screening tests could be performed using the original heelstick sample, without substantial increases in laboratory costs, the cost-effectiveness of newborn screening would be improved.

A critical step in conducting a cost-effectiveness analysis is determining what components are used to estimate the cost of screening. Often studies have

included only the cost of the screening itself, and not all 5 components of the newborn screening system.^{1,14,38,40,96,195,198,200,201} Additional elements that might be included, if the whole system were taken into consideration, are:

- In the screening component—the cost of informing families, obtaining a specimen, and laboratory analysis (including cases that prove to be false-positive);
- In follow-up and diagnosis—the cost of reporting and retrieving results, finding the family, performing a specialty diagnostic evaluation, and identifying the medical home; and
- In long-term management and treatment—the cost of multidisciplinary specialty services, special formulas and foods, durable medical equipment such as hearing aids, and ancillary services such as physical, occupational, or speech-language-hearing therapy.

Equally important to determine is a decision about which costs to include in the estimate of averted costs. For example, there are numerous financial implications associated with a chronically ill child within the context of a family. Unfortunately, because there are insufficient data on some conditions included in newborn screening programs, reliable estimates of averted costs related to these conditions cannot be made.

The OTA recommended that states continue to evaluate the effectiveness and cost-effectiveness of newborn screening programs as new tests become available; particularly the incremental effectiveness of incorporating new tests into various screening strategies (eg, single versus repeat sampling). They also concluded that the federal government “might put as a priority, the collection and evaluation of data that would allow careful analysis in [the] future of costs, as well as effectiveness of widespread screening for these disorders.”¹⁴ As the Human Genome Project moves from basic to applied science, and as this knowledge is incorporated into newborn screening programs, this type of federal investment should be given further consideration.

New Health System Economics: The Era of Managed Care and Integrated Delivery Systems

The health care system has changed dramatically over the past decade and has been shaped by concern about health care cost, and the growth of managed care and its associated changes in the health care delivery system.^{71,133,202} Public and private purchasing dollars have been consolidated, premiums and fees have been curtailed, and patients have been assigned to primary health care professionals/case managers who could act as gatekeepers. Newborn screening systems have been affected by these trends because they operate at the intersection of public health and medical care.^{65,71,72}

The Impact on the Health Care System

The most visible change in the health care system is the shift toward the purchase of managed care

arrangements, and a trend away from traditional indemnity insurance. A managed care organization is an agency through which services are purchased involving a network of health care professionals selected and overseen by the entity.⁷² Typically, the managed care plan (and often its network of providers) assumes financial risk. Managed care organizations have attempted to: 1) organize relationships between health care professionals, 2) limit what would be covered, and 3) control enrollee access to services.⁷² These specifications are defined in contracts between the purchaser and the managed care organization, as well as between the managed care organization and its network health care professionals.

Across the country, the transition of managed care has changed the structure and organization of medical practice. There has been: a shift away from inpatient care, a development of integrated health systems, a reorganization of health provider networks and relationships, a greater emphasis on accountability for cost and quality frequently through shared risk, and increased oversight from federal and state governments. Together, these trends have significantly affected the administrative side of physician practice.⁷¹ Physician and patient relationships have been affected and sometimes disrupted.²⁰³ Physicians have raised concerns about their ability to make referrals to appropriate specialists and subspecialists under third-party payer and managed care controls and restrictions.

The Impact on Public Health Agencies

In the wake of managed care developments, public health agencies and their population-based public health programs have faced fiscal and programmatic challenges. In fiscal terms, agencies with clinic-based services (eg, immunization, sexually transmitted disease testing) have experienced a loss of Medicaid patients and revenues when beneficiaries were assigned to a primary care health professional in private practice. With Medicaid buying managed care arrangements instead of fee-for-service care, the amount of Medicaid dollars available to support public health clinics is reduced.⁷²

However, each public health agency retains programmatic responsibility for population-based programs that protect the public's health.²⁰⁴ Despite decreased fiscal support, health departments have to consider the following:

- What functions and responsibilities must continue to be conducted by public health departments and how best are these public health activities financed?
- How should public health departments interact with managed care organizations and other third-party payers (eg, act as partner, service provider, or regulator)?
- What role should public health departments play in assisting managed care organizations and other third-party payers to integrate preventive medicine and health promotion into their products and services?

- What strategies lead to successful collaboration between public health and managed care organizations/third-party payers?

The Impact on Newborn Screening Programs

For newborn screening systems, public health departments continue to play an essential role in ensuring the service, including financing some aspects of the program with public dollars. For example, states may use tax dollars to supplement fees for newborn testing, to operate a state public health laboratory, to employ staff who do initial follow-up with physicians and families, and to finance treatment for uninsured or underinsured children.²⁰⁵ Public health agencies also have other responsibilities with costs attached, such as monitoring the quality of newborn screening laboratory services, ensuring the completeness of screening and follow-up, operating information systems, and protecting confidentiality and privacy.

In terms of testing, length of hospital stay for the newborn is an issue closely related to newborn screening and its costs. Before 1996, states reported that some newborn screening laws or regulations required repeat screening after early hospital discharge.^{6,206} In 1996, more than half of the states adopted new laws or regulations related to insurance coverage for newborns who are discharged early (typically defined as before 48 hours after a vaginal birth and 96 hours after a cesarean birth), partially in response to concerns about the reliability of newborn screening tests based on samples collected from infants aged 24 hours or less.^{116,207–210} Many of these new laws required that health plans cover 1 or more newborn visits (in the home or clinical setting) subsequent to early hospital discharge that must include collection of an adequate sample for newborn screening (eg, Indiana, Kentucky, Missouri, New Hampshire) or “medically necessary and appropriate tests.” When appropriately implemented, these laws provided for additional payments to cover the cost of repeat testing.

However, repeat testing costs are only one small component of a newborn screening system. As with other population-based public health services that have a medical care component, third-party insurance purchasers and managed care organizations may not recognize the importance of third-party payment for the newborn screening system. Newborn screening services are an accepted and essential component of pediatric care, and should be a service covered and delivered by any third-party payer. Managed care organizations and other third-party payers have a role to play in all parts of newborn screening, including testing, initial follow-up, diagnosis, and management through long-term treatment and follow-up. For example, testing fees may be included in the hospital costs or be a separate cost for a newborn, and the cost of retrieving and reporting newborn screening test results are a part of the cost of initial visits to primary care health professionals. There is little evidence that managed care organizations or other third-party payers have been actively

involved in newborn screening systems. This is an area for further study and improvement.

Maintaining the quality of newborn screening systems amid these changes requires the commitment of public health agencies, health professionals, and managed care organizations/third-party payers. Each third-party payer or managed care organization must have the responsibility to ensure that these services are readily available in the network or by referral to health care professionals and facilities outside the network.²¹¹ Some diagnostic and treatment services needed for follow-up of newborn screening require both expertise available only through a pediatric specialist or subspecialist, and the ongoing comprehensive care that these subspecialists provide. In some cases, appropriate services will only be obtainable outside the third-party payer network through health care professionals or facilities with teams of professionals who specialize in a particular condition. For example, diagnostic services and design of a plan of care might best be achieved through a center of excellence or subspecialty center that has expertise in sickle cell disease, metabolic conditions, or speech-language-hearing treatment of the very young child.

Financing Newborn Screening Systems

States fund newborn screening programs in different ways. Most states set and collect fees for newborn screening tests. However, fees alone are not adequate to finance a newborn screening system, and public health funding is often used to supplement these fees.

Fees for Newborn Screening

States report use of the following funding strategies for newborn screening programs (based on 1996 information submitted by the states to the CORN)⁶:

- Most states billed patients, health care professionals, hospitals, or third-party payers a newborn screening fee. Some states reported no billing and used only public dollars. Eight relied on state general funds (Georgia, Indiana, Kansas, Maryland, New York, North Carolina, Texas, and Wyoming) and 6 used federal grants (Georgia, Kansas, Maryland, New York, Pennsylvania, and Texas).
- Among the 23 states that provided data, the fees charged per newborn ranged from approximately \$40 in Delaware and Massachusetts to less than \$15 in Kentucky, Minnesota, and New Hampshire. These variations reflect both the number of disorders states choose to screen for and the different levels of services supported by newborn screening fees.

A survey on state fees in 1992 showed similar findings, including the following:

- Forty states had set fees for newborn screening and collected them.
- In 21 of the 40 states that charged fees, the laboratory was responsible for fee collection.
- The testing fee included both laboratory and other program services in 30 states. Of these states, 17

financed more comprehensive services including some follow-up and treatment costs.

- While 17 states placed fees into a fund designated for newborn screening program support (and 8 into a special laboratory fund), 10 states returned collected fees back to general revenue budget funds.

Experience of the MCHB technical assistance team to state newborn screening programs indicate additional costs to consumers or their insurers. These include hospital fees that may be sizeable (\$100 in some hospitals) for heelstick blood collection.

Programs Designed for CSHCN

Title V of the Social Security Act mandates that each state put in place community-based, family-centered, culturally competent, coordinated systems of care for CSHCN. Healthy People 2000 called for implementation of these comprehensive systems in all states by the year 2000.

Much has been achieved in establishing these systems, but much remains to be done to accomplish full implementation for all CSHCN. These are defined as “those who have or are at increased risk for a chronic physical, developmental, behavioral, or emotional condition and who also require services of a type or amount beyond that required by children generally.”²¹² Healthy People 2010 reiterates the establishments of these comprehensive community-based systems as a goal, and the HRSA’s MCHB is working actively with the states and other partners to make that goal a reality within the next 10 years.

The federal Supplemental Security Income (SSI) program provides income support and Medicaid coverage to children with disabilities. However, the eligibility definitions of the program have been changed several times in recent years through modifications in federal law or regulation. The result is that families and health care professionals may be confused about the status of children with certain conditions. In addition, reports from pediatric subspecialists suggest that some SSI guidelines for conditions such as sickle cell disease (which is included in state newborn screening programs) do not fit with accepted professional views on the severity of the disease.²¹³

All states have early intervention programs for infants and toddlers, with some covering children diagnosed with certain conditions and others including children “at-risk.” These early intervention programs are administered by a variety of state agencies; one-third are administered by MCH Title V programs. Similar to special education programs, federal rules require states to identify and serve eligible children. Many children with genetic conditions are included in these programs. State education, Medicaid, and public health agencies have administrative and fiscal responsibility for these services.

Health Care Coverage

Health coverage costs are a significant budget and policy issue in every state. Children’s health cover-

age is of particular importance to states, with over one-third of all US births being financed by Medicaid and, in most states, more than half of children using Medicaid or the State Children’s Health Insurance Programs (SCHIP). States have regulatory authority over many insurance practices, both in the public and private sectors. In other words, state actions have substantial influence over whether children with conditions identified through newborn screening have health coverage and how adequate that coverage will be to meet their care and treatment needs.

Private insurance. For children covered by insurance, the Health Insurance Portability and Accountability Act (HIPAA, also known as the Kassebaum-Kennedy legislation) offers protection for newborns in every state.^{214,215} The HIPAA prohibits preexisting condition exclusions for babies if their mother is covered (whether covered by private insurance or Medicaid), and if the infant is enrolled in the plan during the first 60 days of life. In addition, when coverage starts in infancy, the HIPAA provides that prohibitions on preexisting condition exclusions can be effective throughout childhood and beyond. This HIPAA provision was designed specifically to protect infants with genetic, chronic, and other disabling conditions that formerly were considered “preexisting conditions” under many private health plans. However, because states had to conform to many larger provisions of the HIPAA, infant coverage has not been successfully discussed and actively enforced.

States can mandate that benefit packages of private health insurance products include items such as special formula or nutrition supplements. Several states have adopted such mandates. However, because of the Employee Retirement and Income Security Act (ERISA), states’ mandates do not affect employer-based benefit plans that are self-funded (also known as “self-insured”).²¹⁶ As a result, as many as 25% to 50% of those covered under private employer-based plans are estimated to not be protected by the requirements of state insurance benefit mandates.

States have used regulatory authority to direct other types of health plan practices beyond benefits. In terms of managed care, states have adopted a variety of regulations, including some approaches that could be modified to protect CSHCN. For example, by late 1997, 22 states had enacted laws requiring that health plans permit direct access to a particular type of specialist. However, none of these laws specifically addressed direct access to specialty care for children with chronic or disabling conditions. In 18 states, each health plan is required to establish a procedure by which an enrollee may secure a standing referral to a specialist. CSHCN would benefit from this type of protection.

Medicaid. Medicaid is an important source of coverage for children with conditions identified through newborn screening. Medicaid finances an estimated 40% of births, and these infants are automatically eligible as newborns and remain eligible throughout the first year of life. Although federal Medicaid law

requires states to implement automatic newborn eligibility rules and guaranteed coverage for the first 12 months of life, many states do not have effective procedures to implement these guarantees. Medicaid also has a comprehensive benefit package, known as the Early and Periodic Screening, Diagnosis, and Treatment (EPSDT) program. Through the EPSDT program, Medicaid requires that states provide coverage and financing for a wide range of care and services that are medically necessary for CSHCN; including formulas, hearing aids, and therapies needed by children with conditions identified through newborn screening.

Since 1993, most state Medicaid programs have moved toward the purchase of managed care coverage for beneficiaries.⁷² Although few managed care plans have had previous experience providing services to CSHCN, studies suggest that managed care decreases the utilization of specialists by children. One study of Medicaid managed care contracts recommended that state Medicaid contracts: clarify pediatric benefits, define capacity requirements of health care professionals, develop a medical necessity standard specific to children, identify pediatric quality-of-care measures, set pediatric capitation rates, and create incentives for quality in pediatric care.²¹⁷

SCHIP. For low-income children who are not privately insured and not eligible for Medicaid, Congress enacted Title XXI of the Social Security Act, which established the SCHIP in 1997.²¹⁸ Under this program, states are given grants to subsidize health coverage for these children. States may choose to purchase coverage through Medicaid or private insurance (and in some cases directly purchase services from health care professionals). Each state can establish its own guidelines for eligibility based on income, age, disability status, and so forth. If not using Medicaid, states also determine what benefits will be covered under the SCHIP plans. A recent review of the benefit packages of non-Medicaid SCHIP plans found that 5 states do not cover hearing aids, durable medical equipment, and other devices; and that coverage for therapies is uneven with exclusions of developmental conditions or chronic conditions not expected to improve.²¹⁹

Goals for States' Financing of Newborn Screening Systems

In discussions regarding the financing of newborn screening systems, the Task Force identified 3 distinct goals:

- *Adequate financing for screening, short-term follow-up, and diagnosis.* The screening, follow-up, and diagnosis components of the system are generally funded by some combination of newborn screening fees and public dollars. Many states cover most or all of the costs for testing with newborn screening fees; some states supplement or cover screening test costs through general public health funding. Sufficient funds from fees and/or public funds are not always available, however, to ensure adequate short-term follow-up and diagnostic testing. Reliance on third-party payers for short-

term follow-up and for diagnostic testing and interpretation is problematic, in part because these activities need to be conducted expeditiously and because the health insurance status of newborns is often uncertain.

- *Adequate financing for comprehensive care and treatment of all individuals with conditions identified through newborn screening.* Funding for comprehensive medical care and treatment is challenging, and treatment of some conditions identified through newborn screening is costly. Not all children have health coverage or the means to purchase needed treatment. Managed care plans and other third-party payers often do not cover items such as special formulas, special foods, neurodevelopmental assessments, and therapies. Important psychosocial services and other support services for families are also less likely to be funded through health plans. Many managed care plans restrict access to specialized services or require that in-network health professionals who lack appropriate expertise deliver care. For children with complex conditions, treatment may best be delivered by a multidisciplinary team with specialized expertise; however, development and support of such teams requires financing beyond that provided through any form of insurance. Thus, many children with the disorders identified by neonatal screening do not receive optimal care because they have inadequate insurance coverage and/or lack access to qualified health professionals. For many, the situation is exacerbated when they reach adulthood and no longer qualify for public programs such as Medicaid, the SCHIP, and Title V-funded programs for CSHCN.
- *Adequate financing for program evaluation and quality assurance.* Public health agencies and newborn screening program staff are essential to the success of newborn screening systems through their role in activities to ensure laboratory quality, outreach and tracking of families, long-term follow-up, and so forth. State public health agencies and their newborn screening program units should interact with and ensure the quality of all parts of the newborn screening system. Currently, most states do not provide financing for outcomes data collection and evaluation, and this limits their ability to improve the system and to evaluate cost-effectiveness.

Task Force Recommendations to Improve Financing of Newborn Screening Systems

- States should assure adequate financing of all parts of the newborn screening system: screening, short-term follow up, diagnostic testing, comprehensive medical care/treatment, and evaluation of the system. If newborn screening fees are not adequate, funding of all components of the system could be accomplished with other public health dollars or by third-party payers. Other uses of newborn screening fees should not be considered until all of the components of the newborn screening system are fully funded.

- States should take responsibility for blending resources available through Title XIX (Medicaid), Title V (MCH Block Grant), Title XXI (SCHIP), and private insurance to guarantee necessary coverage and financing for all children and adolescents with a condition diagnosed through the newborn screening system.
- State contracts for publicly-subsidized third-party insurance plans that cover children (eg, Medicaid and SCHIP) should explicitly require coverage for newborn screening and those services, including management and treatment, related to disorders identified by newborn screening. State contracts should require that third-party payers ensure access to health care professionals with appropriate pediatric expertise within the network or through out-of-network referrals.
- States, in cooperation with health professionals and payers, should put mechanisms in place to identify the third-party payers for newborns immediately following birth. For example, all states should operationalize the automatic newborn eligibility requirements under Medicaid and the HIPAA newborn coverage provisions that require infant coverage and prohibit preexisting condition exclusions for newborns.
- Purchasers—public and private—should ensure that the benefits packages they pay for includes the care and services defined by the AAP Scope of Health Care Benefits Statement and the CORN guidelines.^{9,143}
- In the SSI program, the federal government should review the technical appropriateness of guidelines, and evaluate the consistency of their application, for children with conditions identified through newborn screening.

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