

Produced by ASTHO in partnership with the Association of Maternal and Child Health Programs
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Introduction

Newborn screening is widely viewed as a great public health accomplishment. Each year, approximately four million infants are screened in the U.S. for a variety of genetic and other congenital disorders. The early identification of infants with congenital disorders is vital to preventing significant morbidity and mortality. State public health agencies have been administering newborn screening programs for more than 40 years. Over the years, changes in technology have contributed to screening advances. To keep abreast of these changes, the public health system must be flexible and continually assess ways in which it can create worthwhile partnerships to provide optimal public health services. Recent challenges in expanding newborn screening include issues related to testing with tandem mass spectrometry and DNA. While the underlying issues are technical in nature, they involve complex systems and capacity issues as well.

Newborn screening is not just a laboratory test. It is a system encompassing five main components: screening, follow up, diagnosis, treatment and management, and evaluation.¹ Education is included throughout the entire system. For the system to function optimally, each of these components must be well coordinated. In addition, adequate funding is needed to support the integrated system, from specimen collection through follow up and evaluation. Failure to consider the system in its entirety can result in a fragmented system unable to effectively and efficiently benefit the public's health.

State public health agencies play a vital role in providing the leadership needed to manage and carry out newborn screening programs by coordinating the system components, ensuring stakeholder involvement, educating health care providers, families and policymakers about the system, and conducting surveillance and monitoring activities to ensure quality. Some state public health agencies also deliver direct health services to families as part of the follow-up and treatment components of the newborn screening system.

State newborn screening programs have functioned and expanded differently over the years as a result of differing state infrastructures and financing mechanisms, as well as varying statutory requirements defining the scope of mandated services. Each state is responsible for designing and implementing its' own newborn screening program, since there has not been federal guidance and oversight. Thus, each system has evolved in the context of the individual state's economic, demographic, geographic, and political manifestations. While this allows flexibility for state newborn screening programs, it has also led to significant variation among programs with the result that access to newborn screening services vary from state to state. This variation has drawn concern from many groups; currently there are a few national initiatives examining the issues involved in recommending a uniform panel of tests for states.

This issue brief provides an overview of the newborn screening system and highlights ways state public health agencies can support a coordinated, seamless system that benefits infants, families and providers. This brief focuses primarily on state-level actions. Federal

leadership and support for each of these components is also critical.

The Newborn Screening System

Newborn screening is a complex public health system that saves lives and improves health outcomes. According to the General Accounting Office (GAO), states spent over \$120 million on newborn screening in 2001. Laboratory expenditures accounted for 74 percent of states' expenses, with the remaining 26 percent going toward program administration and follow-up.ⁱⁱ The screening, education, diagnosis, and follow-up components of the newborn screening system are generally funded by some combination of newborn screening fees and public dollars, although at least eight states do not charge a fee for newborn screening. Medicaid, third party payers, and the Maternal and Child Health (MCH) Block Grant also contribute to the newborn screening system.

The mechanism for funding newborn screening can lead to fragmentation, because funding allocations for different parts of the newborn screening system may be uneven or overlooked. Where they exist, newborn screening fees are usually attached to laboratory services and may not consider the costs of other system components. Extracting laboratory testing from the overall newborn screening system in order to produce a lower cost test could result in a fragmented and inefficient system if sufficient fees are not appropriated to continue to pay for the newborn screening system. Insufficient funding can lead to incomplete follow up of abnormal or unsatisfactory testing results, variable testing quality, fragmented data for program evaluation, and incomplete coverage of those who are unable to pay for newborn screening services.

Screening

Since the early 1960s, newborn screening has been a public health activity generally housed

within the state public health agency. Budgetary constraints and demographics (e.g., the number of births in a state) have contributed to decisions related to direct newborn screening service delivery.

Over the years, delivery of newborn screening laboratory services has resulted in several different models, including: 1) a single state public health laboratory or university laboratory; 2) a regional public health contract; 3) a regional private contract; and 4) singular or multiple private laboratories under state contract. The first model continues to be the most common arrangement used. The decision to provide a service directly or through a partner primarily relates to laboratory testing, but in some cases includes the follow-up and education services.

Contracting with other state labs or private entities to perform the state's mandated panel may bring significant benefits to the state, including cost savings and quicker analysis of tests. However, states face increasing challenges posed by private laboratory services being offered in some areas as an alternative to the state mandated program; in some cases private laboratory services may be offered in competition with the state's mandated program. Without certain policies in place, these arrangements may lead to fragmentation of the system through questions over coordination and each entity's responsibilities, and create a need for new policies and communication systems. Thus, state public health agencies must develop and sustain relationships with public and private entities that integrate screening as just one component of the system.

To promote coordination in screening activities, state public health agencies can:

- Work with state medical associations, primary care associations, and others to develop and provide ongoing training around state mandates, procedures in newborn screening, and advances in technology. State public health agencies can create and disseminate newborn screening manuals to serve as a reference on procedures and each entity's responsibilities. States can also work with

residency programs to hold “grand rounds” on newborn screening, particularly when the panel changes or new technology is introduced. Some state public health agencies use the media to educate providers and the public, for instance submitting articles to the state medical journal.

- Educate stakeholders so they understand the options available to families around newborn screening testing, including tests that may exist beyond the mandated program.
- Partner with families to design educational materials that inform families about newborn screening during the prenatal period. State public health agencies can work with parent groups to design and test materials and different means of communication to families. State public health agencies should ensure that materials can reach families with low literacy levels and of different cultures. Some states design brochures, posters and videos for use in prenatal care provider offices and parenting classes.
- Work with prenatal and pediatric care providers to disseminate materials to families. State public health agencies can also work with hospital neonatal and discharge teams to provide or reinforce education to families at the time of testing.
- Educate contracted labs on how newborn screening fits into the overall newborn screening system and jointly develop educational materials to disseminate to families and providers with the screening test kit. Private labs may assume that all providers and families are aware of the entire expanded screening process.
- Explore the feasibility of adopting a single form that can be used for both the mandated and expanded screen. Some states that offer expanded screening outside of the public health lab are considering using a single form with two filter papers to simplify the processing of results for mandated and expanded specimens.

Follow Up

Rapid follow up of an infant who tests positive is vital to reducing morbidity and mortality and requires timely communication of the results to the hospital, primary care provider and family. Both short and long term follow up are critical components of the newborn screening system. Short-term follow up generally encompasses the time from when a test is suspected as positive until the infant receives appropriate treatment, while long-term follow up consists of ongoing treatment and monitoring of children. This section refers to short-term follow-up.

There is wide variation in states’ capacity to conduct follow up for newborn screening. Follow up activities may be comprised of telephone calls and letters to providers and families, as well as home visits or case management, depending on the size and budget of the program. In general, state public health agencies have a role in assuring that primary care providers receive newborn screening results. In some states the lab has responsibility for notifying the health professional and/or family, and in other states this is the responsibility of the Title V agency. Because contracted labs are not typically responsible for follow-up activities, state public health agencies must work with them to ensure that appropriate communication occurs.

The availability of expanded screening by the private sector may present additional challenges to states around follow up. Children identified with a disorder through expanded programs are referred to the state public health agency for services. Yet, state public health agencies may not have funding to provide follow-up services for children identified outside of the mandated program or have a system set up to track those children testing positive through the private sector not mandated by the state. These situations not only add financial burden to the states, but also increase liability.

To improve coordination around follow-up activities, state public health agencies can:

- Facilitate relationships between primary care providers, pediatric subspecialists, state

medical associations, and other service providers to develop and provide ongoing training around follow-up procedures for newborn screening and each agency's or stakeholder's responsibility for insuring coordination of the system. State public health agencies can help develop training and resources on the provider's role in linking families to specialty care. State public health agencies can also explore opportunities to work with those partners to provide "enabling services" such as care coordination to families. Some state Title V programs work with the contracted lab to track the infant until a definitive diagnosis is determined.

- Work with hospital obstetric and discharge teams to ensure that a primary care provider (or medical home) is identified for all infants at the time of the screen to help simplify communication of test results to the provider and facilitate the follow-up process.
- Develop communication plans with labs to ensure that all results, whether positive or negative, are shared with the primary care provider. Sometimes providers do not receive all results and assume the infant has tested negative. State public health agencies can also create a central point of contact at the lab, whether it is the state lab or a private lab that they can contact with questions to expedite follow up and improve coordination.
- Ensure that contracts stipulate that information be provided to primary care providers and the state public health agency to facilitate appropriate follow-up activities. Some states have developed mandates requiring that the private lab return some portion of the fee collected to the state to use for short-term follow-up activities.
- Support primary care physicians by providing timely information on a patient's condition after a positive test, as well as information on appropriate follow up to share with families. Some state public health agencies call and fax the primary care provider when a patient screens positively

for a genetic or metabolic condition and provide information on the disorder, as well as recommendations for follow-up and information on metabolic specialists in the state.

Treatment and Management

State public health agencies typically have a role in linking families to services after a diagnosis is confirmed. Administering a follow-up system is a costly activity and few states have the resources to conduct comprehensive, long-term follow-up. States also differ in the type of follow-up performed, ranging from tracking the infant only through diagnosis and initial treatment, to following children with certain conditions until 21 years of age. Extensive long-term follow-up can allow states to monitor children and create a registry of information to evaluate services and the newborn screening system.

Adequately financing the treatment of children identified with genetic conditions can also be difficult for states. Families face significant financial burdens accessing treatments needed to prevent serious disability or death. Some insurers do not cover necessary treatments such as the special foods and formula needed for children with phenylketonuria (PKU), creating great financial strain on families. Without special diets, children are put at risk for severe health problems. Many Title V agencies assist with these costs, however the aid ends either at age 18 or 21.

To improve the treatment and management of infants with genetic or metabolic disorders, state public health agencies can:

- Fund a nurse coordinator or case manager to help families access and understand services. Some states fund nurse practitioners or other providers to work as a coordinator for families at metabolic clinics to help families access and understand needed services. At least one state established regional "child health teams" made up of a social worker and a nurse that perform follow-up services to infants who screen positive as part of their larger child health

responsibilities such as home visits and immunizations.

- Ensure that a portion of the screening fee is directed to the state public health agency to be used for follow-up activities, such as case management. State public health agencies can identify this funding separately from the laboratory contract to assure an ongoing funding stream.
- Promote state-based information integration initiatives to share information among the state public health agency, primary care providers, the lab, and others. Several states are integrating infant data from vital statistics, newborn screening, birth defect registries, immunization registries and other child health programs to create a child health profile. These systems can help to improve coordination of services, facilitate communication and improve monitoring of infants.
- Work with insurers to cover special formula and nutritional foods. States can also work with insurers to raise annual caps on reimbursement of formulas and special foods, as well as restrictions for individuals over 18 years and who no longer are eligible for certain public programs.

Evaluation and Quality Assurance

Ideally, state public health agencies will gather information on the performance of the entire newborn screening system, including monitoring screening coverage, follow-up rates, and health outcomes, as well as assessing stakeholder satisfaction with the system. However, resources are often insufficient to fund comprehensive quality assurance and evaluation activities, and so they often focus on the lab activities. Each lab's performance must be monitored for quality and cost-efficiency and have adequate quality control to ensure maximum disease detection with minimal false negative results.ⁱⁱⁱ All states participate in quality assurance activities. Other state public health agency evaluation activities include

collecting data to inform state policymakers about which tests to include in panel, to develop the evidence base for state decisions, and for research to determine the effectiveness of new technologies. Some states have legislation to monitor compliance with state laws around newborn screening.

Other important quality measures focus on protection of families and ethical handling of dried blood spots. New technologies have led to the need to develop policies on patient confidentiality, ownership and storage of specimens, particularly those collected through expanded screening programs. These developments present new ethical challenges to the newborn screening community and require an active leadership role by the state public health agency.

To enhance evaluation and quality assurance activities, state public health agencies can:

- Ensure that a variety of stakeholders, such as pediatricians, obstetricians, hospital administrators, families, public health and laboratory staff, and insurance companies is represented on the state newborn screening advisory board. Some states have both internal and external advisory committees where the external group makes recommendations to the internal group of state health officials. This separation can help prevent potential conflict of interest for individuals on the external group who may be involved in newborn screening research activities.
- Be involved in quality assurance of services that the state public health agency does not directly provide. For instance, states can include reporting requirements and quality control activities in contract specifications with private labs.
- Share policies on privacy, ethics, and appropriate consent for the ownership and use of specimens with stakeholders to ensure optimal protections for families. State public health agencies can work with private entities to create and enforce policies that promote privacy for families and that address consent

issues for research and future use of the testing sample as needed.

- Educate stakeholders on confidentiality and protection issues in research and storage of samples, as well as use of patient data. State public health agencies can work with stakeholders to develop and provide training and other information on these issues.
- Participate in national training and quality assurance activities, such as the Newborn Screening Quality Assurance Program and other training offered through APHL and newborn screening equipment manufacturers. State public health agencies can also request a technical review from the National Newborn Screening and Genetic Resource Center to help create procedures for evaluation and quality assurance.
- Use data from the four MCH performance measures that relate to newborn screening to contribute to program evaluation and emphasize system needs. States can also establish new measures under state negotiated performance measures. States can disseminate this data to policymakers and stakeholders to demonstrate success and areas of need.

Conclusion

Advances in technology, combined with economic, geographic and political demands, have changed the environment in which newborn screening programs operate. As state newborn screening programs continue to expand, state public health agencies are faced with new challenges and opportunities related to ensuring effective, seamless, and ethical newborn screening programs. As a result, state public health agencies must adopt new policies and partnerships to ensure that all newborns receive timely and appropriate testing and follow up for genetic and other congenital disorders.

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ⁱⁱ United States General Accounting Office. Newborn Screening. Characteristics of State Programs. March 2003.

ⁱⁱⁱ Therell BL, et al. U.S. Newborn Screening System Guidelines: Statement of the Council of Regional Networks for Genetic Services. February 1992.