

Examination of the Communication Practices Between State Newborn Screening Programs and the Medical Home

Sunnah Kim, MS, RN, CPNP*; Michele A. Lloyd-Puryear, MD, PhD, FAAP‡; and Thomas F. Tonniges, MD, FAAP*

ABSTRACT. *Background.* As outlined in the Newborn Screening Task Force report published in August 2000, the newborn screening system is more than just testing, but also involves follow-up, diagnosis, treatment, and evaluation. As such, multiple professional and public partners need to be adequately involved in the system to help ensure success. In addition, newborn screening programs are state-based; therefore, policies and procedures vary from state to state. Historically, there has been little uniformity between state newborn screening programs.

Objective. To examine the communication practices of state newborn screening programs in the United States, particularly in relation to the medical home.

Methods. A facsimile survey of program staff in all US newborn screening programs. Survey data were collected in August 2000.

Results. All 51 programs participated. States were questioned about whether or not they had a procedure to identify the infant's medical home before the child's birth. Twelve states (24%) indicated that there was a procedure in place, whereas 39 states (76%) indicated that either no procedure existed or that they were unsure. In contrast, all state programs (except 1) indicated they notified the primary care physician about abnormal results and the need for follow-up. In addition, state programs reported that primary care physicians have responsibilities within the newborn screening system, particularly related to communicating with parents about screen-positive results and coordinating the collection of a second specimen. Thirty states reported that they directly notified parents of screen-positive infants of results and the need for follow-up as well.

In regard to informing parents about newborn screening, 45% of states indicated that primary care physicians had some responsibility in informing parents about newborn screening. Most often, parents were informed about newborn screening just before specimen collection, and the most commonly used techniques to educate parents were informational brochures and conversation.

Thirty-five states reported that they engaged in long-term tracking of infants after diagnosis confirmation. Only about half of these states provided long-term tracking of all of the conditions included in their state's newborn screening test panel. Of these 35 states that engaged in long-term tracking, 25 reported that they requested patient information from the primary care physician and/or subspecialist about ongoing treatment and follow-up.

Conclusions. Newborn screening roles and responsibilities vary tremendously between states. Improvements in communication and better-defined protocols are needed, particularly between state newborn screening programs and the medical home. Many states identified the medical home as having significant responsibilities related to the short-term follow-up of screen-positive infants. Identification of the correct medical home before testing would help to reduce unnecessary time and frustration for state newborn screening programs, especially in the follow-up of infants that are difficult to locate. In addition, primary care physicians (ie, the medical home) need to have appropriate and ongoing involvement, including a mechanism to provide feedback to their state newborn screening program. This is particularly important given the adoption of tandem mass spectrometry by an increasing number of states, and the likely expansion of newborn screening in the future. Recommendations include the following:

- Primary care physicians should have appropriate and ongoing involvement in the newborn screening system and should be appropriately represented on state newborn screening advisory committees,
 - States should develop protocols to identify the medical home before heelstick screening,
 - States should work with families, primary care physicians, and prenatal health care professionals to develop well-defined systems for pretesting education of parents,
 - All newborn screening results (both positive and negative) should be sent to the infant's medical home. If results are not received by the medical home, efforts should be made to obtain results,
 - Medical homes and subspecialists should submit follow-up information on screen-positive infants and infants with confirmed diagnoses to the state newborn screening program, regardless of the existence of state requirements to do so, and
 - Efforts to build enhanced direct communication systems, linking state newborn screening programs to community-based medical homes, should continue.
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From the *Department of Community Pediatrics, American Academy of Pediatrics, Elk Grove Village, Illinois; and ‡Genetic Services Branch, Maternal and Child Health Bureau-Health Resources and Services Administration, Rockville, Maryland.

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Reprint requests to (S.K.) Department of Community Pediatrics, American Academy of Pediatrics, 141 Northwest Point Blvd, Elk Grove Village, IL 60007-1098.

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State newborn screening programs have existed for the past 35 years and are considered one of the greatest accomplishments of public health.¹ Each day, newborns across the country receive screening for metabolic, genetic, infectious, and other congenital conditions that threaten their health and life. Results most often indicate that screened newborns are not likely to have these life-threatening disorders; most infants screen negative (normal). However, in cases of positive (abnormal) results, much needs to be done to ensure appropriate follow-up of screen-positive infants, early diagnosis confirmation, and treatment. Prompt action is especially needed for certain disorders (eg, galactosemia, congenital adrenal hyperplasia), as delay in treatment leads to increased morbidity and mortality.

As stated in the Newborn Screening Task Force Report, "Serving the Family From Birth to the Medical Home,"¹ newborn screening is more than just testing. Newborn screening is a system involving initial screening, immediate follow-up evaluation of the screen-positive newborn, diagnosis confirmation, initial and long-term treatment and management, and evaluation of all components of the system.^{2,3} For the system to operate effectively, all components must function well and be appropriately interconnected. In particular, a seamless mechanism for communication between the public health system and the medical home needs to exist.⁴

The medical home is defined as care that is accessible, continuous, comprehensive, family-centered, coordinated, compassionate, and culturally effective. This care is delivered or directed by well-trained physicians, in partnership with families, to provide primary care and to help manage and facilitate essentially all aspects of pediatric care.⁴ Most often the medical home is provided by primary care physicians; however, in a limited number of cases, specialty clinics can provide a full range of services (including primary care) and be considered the medical home for a child with complex health care needs. For a child who screens positive, the medical home can play an important role to assist families in scheduling diagnostic testing, to ensure that diagnostic test results are received and shared with the newborn screening program, and to provide the family with appropriate referrals in the event of confirmatory results. The need for a seamless system between the newborn screening program and the medical home is particularly apparent with recent advances in technology such as tandem mass spectrometry and its potential for expanded newborn screening.

However, a seamless system is not always the case, as illustrated in a study by Desposito et al.⁵ This study indicated that 31% of pediatricians were notified of screen-positive results >10 days after testing was completed. Although the majority of these patients received treatment and follow-up by the birth hospital or the state newborn screening follow-up team in a timely manner, communication with the primary care physician was less efficient than recommended guidelines.^{2,3} Communication of screen-negative results was also less than optimal, as 26% of

pediatricians were not routinely notified of results. Although most pediatricians surveyed made an effort to track down the missing results of newborns in their practice, 28% of pediatricians interpreted the lack of a report as being a negative result. This finding provides support to a recent study by Mandl et al,⁶ which indicated that approximately one third of states did not provide a written copy of newborn screening results to the infant's physician.

To further examine the newborn screening system, a survey was mailed to all state newborn screening programs in the United States to better ascertain how the state programs define the roles and responsibilities for the community-based primary care physician and the newborn screening program staff in each respective state. The specific aims of this survey were to: 1) better understand state practices of linking screened infants with the correct medical home; 2) identify areas of the newborn screening system that are collectively viewed by state newborn screening programs as being the responsibility of the primary care physician, and 3) assess the communication practices of state newborn screening programs to community-based primary care physicians as a whole.

METHODS

The 27-question survey tool was developed and pretested during informal discussions with primary care physicians, state public health officials, newborn screening program staff, and other experts. Using a contact list from the National Newborn Screening and Genetics Resource Center, the survey was sent via facsimile in August 2000 to newborn screening program staff in all 50 states and the District of Columbia. A cover memorandum, indicating that the survey was part of a larger project being conducted by the American Academy of Pediatrics and the federal Maternal and Child Health Bureau, was included. The memorandum instructed the facsimile recipient to forward the survey to the person best equipped to answer questions about newborn screening follow-up, if they were not familiar with the subject area. To help eliminate any confusion over terminology used in the survey, a list of definitions was included.

Two months after the initial facsimile distribution, a reminder was sent to the states that had not responded. Individual contact by telephone and/or e-mail was made to the states that had failed to respond to the reminder.

Survey responses were tabulated and organized in a Statistical Program for Social Sciences (SPSS, SPSS Inc, Chicago, IL) database. Descriptive statistics and frequencies were analyzed.

RESULTS

All 51 surveys were returned within 4 months of initial distribution. In all cases, survey responses were provided by a staff member of the state newborn screening program. Responses to key questions are summarized in Table 1.

Pretesting

States were asked, "Is there a procedure for linking all prospective parents with a pediatric primary care physician (before the child's birth) for the purpose of assuring a medical home?" Although a definition of "medical home" was provided in the survey, no definition of "procedure for ensuring a medical home" was given. Twelve states (24%) indicated that there was a procedure for ensuring a medical home, whereas 22 states (43%) stated that no such proce-

TABLE 1. Key State Responses*

	Procedure for Assuring a Medical Home Exists	Medical Home Has Role in Pretesting Education	Directly Notify Parents of Screen-Positive Results	Request Patient Info From Medical Home for Short-Term Follow-up	Engage in Long-Term Tracking	Offers Professional Education and Training	Targets Medical Homes for Education/ Training
Alabama	•	•	•	•	•	•	•
Alaska	?	•		•	•	•	•
Arizona			•	•		•	•
Arkansas	?		•	•	•	•	•
California	•	•	•	•		•	•
Colorado				•		•	
Connecticut			•	•		•	
DC			•	•		•	
Delaware	•		•	•	•	•	•
Florida			•	•		•	•
Georgia		•		•	•		NA
Hawaii	•	•	•	•	•	•	•
Idaho	?	•		•	•	•	
Illinois		•		•	•	•	•
Indiana		•		•	•	•	•
Iowa	?	•		•	•	•	•
Kansas	•	•		•	•	•	•
Kentucky			•	•	•		NA
Louisiana	•	•	•	•	•	•	•
Maine	?	•		•	•	•	•
Maryland			•	•	•	•	•
Massachusetts	?		•	•	?	•	•
Michigan			•	•	•	•	•
Minnesota	•	•	•	•	•		NA
Mississippi			•		•	•	•
Missouri	?			•		•	
Montana	•		•	•	•	•	
Nebraska	?		•	•		•	•
Nevada	•	•		•	•	•	
New Hampshire				•	•		NA
New Jersey	•		•	•	•	•	•
New Mexico	•	•	•	•	•	•	•
New York				•		•	•
North Carolina	?	•	•	•	•	•	
North Dakota	?			•	•	•	
Ohio	?			•		•	•
Oklahoma		•	•	•	•	•	•
Oregon	?	•		•		•	•
Pennsylvania				•	•		NA
Rhode Island	?		•				NA
South Carolina	?	•		•	•	•	•
South Dakota	?			•		•	
Tennessee	•	•	•	•	•	•	•
Texas	?		•	•	•	•	•
Utah				•	•	•	•
Vermont		•		•	•	•	•
Virginia		•	•	•	•	•	•
Washington			•	•	•	•	•
West Virginia		•			•	•	•
Wisconsin				•		•	•
Wyoming	?			•			NA

NA indicates not applicable.

* Dot indicates a "yes" response. Question mark indicates a "don't know" response.

cedure existed. The remaining 17 states (33%) were unsure.

States reported vastly different procedures for informing parents about newborn screening. Most states used informational brochures and conversation and reported that a combination of health professionals shared responsibility in conveying this information to parents. Twenty-three states (45%) indicated that primary care physicians (ie, the medical home) had some responsibility in informing parents about newborn screening. Of these, 1 state (Vermont) identified the primary care physician/medical home as having sole responsibility.

Thirteen states (25%) reported that the state has a policy encouraging or requiring that parents be informed about newborn screening during the prenatal period, as is recommended by the Newborn Screening Task Force.¹ Most commonly, states reported that parents were informed just before specimen collection. Of interest, 3 states were unable to report a procedure for informing parents about newborn screening before testing.

Short-Term Follow-up

After specimen collection and laboratory analysis, effective communication between the state newborn

screening program, the medical home, the subspecialist, and families needs to occur to ensure timely follow-up and diagnosis of screen-positive infants. All but 1 state indicated that they notified the primary care physician/medical home about abnormal screening results and the need for follow-up. In this state (Mississippi), it was reported that results were sent to the hospital of birth. Forty-seven states (92%) requested patient information from the primary care physician/medical home during follow-up. In addition to notifying primary care physicians, 30 states (59%) directly notified parents of screen-positive infants as well. No data were collected about the role of the subspecialist in receiving or communicating abnormal screening results.

Tracking screen-positive infants is critical to ensuring that they are not lost during the follow-up process. All survey respondents indicated that tracking screen-positive results was a state responsibility. In 9 states, this responsibility was shared with primary care physicians. Most states (94%) reported that in addition to tracking screen-positive results, it was also a state responsibility to track negative newborn screening results.

State newborn screening program staff were questioned about roles and responsibilities after initial testing. Table 2 summarizes primary care physician responsibilities in the short-term follow-up of newborn screening results. Approximately half of the states relied on primary care physicians to assist in communicating with parents and in collecting repeat specimens.

Long-Term Tracking

After diagnosis confirmation, 35 states (69%) track positively identified infants over an extended period of time (ie, long-term tracking). About half of these states provide long-term tracking for all conditions that are included in their state newborn screening panel, whereas the remainder of the states provide this service for select disorders only. Of these 35 states, 25 reported that they request patient information and reports from primary care physicians

and/or subspecialists about ongoing treatment and follow-up. Two states requested information from primary care physicians/medical homes, 3 states requested information from subspecialists, and 20 states requested information from both.

Education

States were asked whether or not they provide professional education and training on newborn screening. Of the 44 states (86%) that provided this service, 35 states (69%) targeted the primary care physician/medical home and ancillary staff in their educational efforts. Forty-one states (80%) included the topic of health professional responsibilities in the newborn screening system.

DISCUSSION

Newborn screening for disorders that potentially lead to catastrophic health consequences has been a concern of public health departments, primary care physicians, subspecialists, and families since the development of screening systems in the early 1960s. Within these systems, there are critical intersections between system components where responsibilities and coordinated activities must be seamless and nonduplicative. It is during these intersections that the importance of effective partnerships between public programs and primary care physicians is most evident. Although the benefits of such partnerships are undeniable, little has been written about the relationship between state newborn screening programs and the medical home. In addition, 1 recommendation of the Newborn Screening Task Force was to better define roles and responsibilities in state newborn screening systems.¹

Our survey indicated that newborn screening roles and responsibilities vary between states and do not always include primary care physicians. One example of this state-to-state variability was in the methods used for pretesting education. Generally, the process of informing parents was left to informational brochures that were most often shared just before testing. Primary care physicians were responsible for pretesting education in some states. The absence of a policy, which sets standards and identifies responsibility for informing parents about newborn screening, also compromises the ability to assure that parents have the opportunity, when legally permitted, to refuse testing. A better-defined process for pretesting education, including prenatal education, would help to create the necessary structure within the newborn screening program.

This survey also revealed inconsistencies in the survey responses. For example, almost all states notified the primary care physician/medical home about screen-positive results and the need for follow-up. This finding supports previous studies that indicated that the process for testing and short-term follow-up of screen-positive infants is well-defined by most programs both programmatically and in practice.^{4,6} However, only 24% of states indicated that they had a procedure to ensure that infants were linked to a medical home. Because most states had

TABLE 2. Number of States That Assigned Partial Responsibility for Each Identified Role to the Primary Care Physician/Medical Home

Role	Number of Respondents	%
Notifying parents for suspect results	27	52
Notifying parents for unacceptable specimens	23	45
Notifying parents of infants discharged early	22	43
Collecting second specimen if abnormal results	34	67
Collecting second specimen for unacceptable specimen	27	53
Collecting second specimen for infants discharged early	28	55
Collecting second specimen for transfused infants	31	61
Collecting second specimen for transferred infants	14	28

no mechanism in place to identify the infant's medical home, it cannot always be guaranteed that communication of screening results and the need for follow-up is conveyed to the correct medical home in these states. Although identification of the medical home before specimen collection would simplify the communication of results to primary care physicians and facilitate the follow-up process for the public health system, according to this survey most states were not using this opportunity. By ensuring that all newborns have an identified medical home before hospital discharge, state newborn screening programs also can contribute to the greater success of Medicaid and State Children's Health Insurance Program enrollment activities, a priority of the American Academy of Pediatrics and the federal government.

Improvements to the current short-term follow-up system are needed as studies indicate that pediatricians were notified of screen-positive results within 10 days of testing,⁴ whereas the national recommendation is for primary care physicians to be notified within 5 to 7 days of testing.^{2,3} As most of our survey respondents indicated that short-term tracking of all newborn screening results (both positive and negative) was a state responsibility, newborn screening programs need to ensure more timely and consistent communication with primary care physicians. When results are not received, the medical home should make efforts to promptly retrieve all missing results from the newborn screening program. In addition, any follow-up information on screen-positive infants should be reported to the newborn screening program, regardless of the existence of state requirements to do so. An enhanced direct communication system would help to address this issue and would be welcomed by primary care physicians.⁴

Preliminary efforts are underway to build integrated information systems that would serve as a direct communication system between state newborn screening programs and the medical home. The Genetic Services Branch of the Maternal and Child Health Bureau, Health Resources and Services Administration has funded several states (ie, the District of Columbia, Hawaii, Iowa, Massachusetts, Michigan, Missouri, Oklahoma, Rhode Island, Washington, and Utah) to integrate newborn screening programs, including their information systems, with other child health programs (eg, immunization registries, birth defects registries, hearing screening programs, Women, Infants, and Children, etc) and community systems of care. Challenges include legislative restrictions, the Health Insurance Portability and Accountability Act, funding, technology readiness of the community providers, political will, resources (technical and personnel), and a unified vision. Such integrated information systems are one mechanism that might be used in the future to facilitate communication between state newborn screening systems and community-based primary care physicians.

In addition to short-term tracking and follow-up, long-term tracking has potential benefits for children with conditions identified through newborn screen-

ing. States that monitor these children have a valuable registry of information that can be used to disseminate information about new treatments or other advances in medical knowledge. In addition, long-term tracking has benefits to the newborn screening program itself. States would have the ability to monitor the clinical progress of children, including the effectiveness of treatment, particularly in lesser-known conditions that are added to newborn screening test panels. Data collected through long-term tracking may help states to objectively evaluate their screening program and help determine whether screening should continue for newly added conditions. Interestingly, of the states that were engaged in long-term tracking, about half did not provide this service for all of the disorders that they included in their respective screening test panels. If states choose to include a certain set of disorders in their screening program, it is questionable as to why they would not include the same disorders in their long-term tracking efforts. Most often, time and financial resources are identified as barriers. However, states should consider long-term tracking a necessary component of the newborn screening system.

Most state programs (80%) were involved in educating primary care physicians about their roles in the newborn screening system. These educational efforts are particularly important when changes are made to the state newborn screening program. States should also strive to involve families and primary care physicians in planning these educational initiatives.

This type of survey lends itself to possible limitations. Surveys were completed by a newborn screening staff person identified by the National Newborn Screening and Genetics Resource Center as the person most knowledgeable about newborn screening follow-up issues in each state. Responses were not validated by a secondary source or through other means, although "don't know" was listed as an option for most questions to help minimize incorrect responses. The survey was completed by state newborn screening program staff in August 2000. Policies and practices of the state newborn screening program may have changed since the time of data collection. Secondly, although most of the terminology used in the survey were defined, some were not (ie, procedure for ensuring a medical home). In addition, there was no mechanism to assess whether survey respondents had a good understanding of these definitions (eg, the medical home). Because this survey was designed to examine the relationship between the state newborn screening program and the medical home, a limited amount of data were collected on the role of the sub-specialist within the newborn screening system. This may have implications for some of the conclusions; as it is difficult to judge whether 2 parts of a system interact with maximum efficacy if the role of a third is not fully explored.

CONCLUSION

It is clear from this study that there is a need for improved communication and better-defined proto-

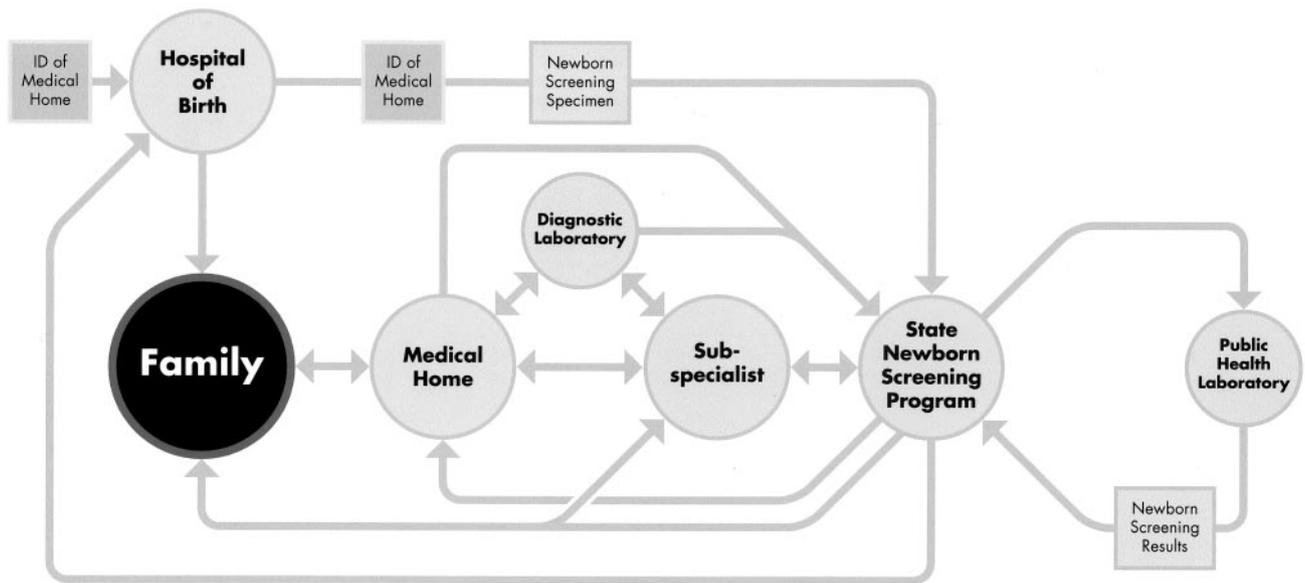


Fig 1. For ideal flow of communication within a state newborn screening system, the medical home should be identified in the prenatal period. If not identified before birth, the birth hospital should ensure that the correct medical home is identified before specimen collection. After specimen collection, the sample is transported to the state newborn screening program, where the sample undergoes laboratory analysis. All newborn screening results should be communicated to the medical home, including all screen-negative results. The medical home should contact the state newborn screening program if screen-negative results are not received. For screen-positive results, the newborn screening program staff should notify the medical home and/or subspecialists, families, and the hospital of birth to ensure timely and appropriate follow-up and diagnosis. The medical home, subspecialist, and state newborn screening program have bidirectional communication, particularly in sharing information on screen-positive infants (ie, test results, clinical information) and in infants who are positively identified and require long-term tracking and follow-up.

cols in state newborn screening programs, particularly with the prospect of expanded newborn screening as the norm. Currently, 20 states utilize tandem mass spectrometry, with many other states considering this possibility.⁷ Appropriate and ongoing involvement of primary care physicians (ie, the medical home) would facilitate state follow-up and tracking efforts (Fig 1). Given the results of this survey and understanding the limitations noted above, we can draw the following conclusions and recommendations:

1. Primary care physicians play an important role in the success of the newborn screening system and should have appropriate ongoing involvement in follow-up and tracking efforts. In addition, primary care physicians should be appropriately represented on state newborn screening advisory committees.
2. States should make efforts to identify the medical home before conducting heelstick testing. States should also utilize this opportunity to enroll eligible infants in Medicaid and State Children's Health Insurance Program.
3. States should work with families, primary care physicians, and prenatal health care professionals to develop a well-defined system for pretesting education of parents.
4. All newborn screening results (both positive and negative) should be sent to the infant's medical home. If results are not received by the medical home, the primary care physician should make efforts to obtain results.
5. Medical homes and subspecialists should submit

follow-up information on screen-positive infants and infants with confirmed diagnoses to the state newborn screening program, regardless of the existence of state requirements.

6. Efforts to build enhanced direct communication systems, linking state newborn screening programs to community-based primary care offices, should continue. Such systems would help to enhance timely follow-up for screen-positive infants and facilitate information sharing among those involved in caring for children.

The success of the newborn screening system depends on effective collaboration and communication between state programs, medical homes, and other components of the newborn screening system. Although some parts of the system currently function well, there are several areas where improvements can be made. These improvements will ultimately benefit the 4 million infants screened each year and assist states in developing the infrastructure that will be needed as expanded newborn screening is ultimately adopted.

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