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Newborn Screening Expands: Recommendations for Pediatricians and Medical Homes Implications for the System

Newborn Screening Authoring Committee

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American Academy of Pediatrics
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CLINICAL REPORT

Newborn Screening Expands: Recommendations for Pediatricians and Medical Homes—Implications for the System

Guidance for the Clinician in Rendering Pediatric Care

Newborn Screening Authoring Committee

ABSTRACT

Advances in newborn screening technology, coupled with recent advances in the diagnosis and treatment of rare but serious congenital conditions that affect newborn infants, provide increased opportunities for positively affecting the lives of children and their families. These advantages also pose new challenges to primary care pediatricians, both educationally and in response to the management of affected infants. Primary care pediatricians require immediate access to clinical and diagnostic information and guidance and have a proactive role to play in supporting the performance of the newborn screening system. Primary care pediatricians must develop office policies and procedures to ensure that newborn screening is conducted and that results are transmitted to them in a timely fashion; they must also develop strategies to use should these systems fail. In addition, collaboration with local, state, and national partners is essential for promoting actions and policies that will optimize the function of the newborn screening systems and ensure that families receive the full benefit of them.

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Key Words
newborn screening, genetic disorders, children with special health care needs, medical home

Abbreviations
AAP—American Academy of Pediatrics
ACMG—American College of Medical Genetics
ACHDGDNC—Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children
MCHB—Maternal Child Health Bureau
HRSA—Health Resources and Services Administration
PCP—primary care pediatrician
CPT—*Current Procedural Terminology*
ACOG—American College of Obstetricians and Gynecologists
SNSAC—state newborn screening advisory committee

INTRODUCTION

> It's another busy day in pediatric practice, even before you receive the telephone call from the state newborn screening program. One of your newborn patients has an out-of-range result* on the screen for a rare but serious congenital condition. "Now what?" you wonder, as you begin to take down the notes. What additional testing is needed? What is the treatment regimen, and when does it begin? What do you tell the parents? And, what do you do about the rest of your schedule?

In the past decade, new technologies have led to a rapid expansion in the number of congenital conditions that are targeted in state newborn screening programs. As newborn screening programs expand, the likelihood increases that individual pediatricians will one day receive an out-of-range screening result for an unfamiliar congenital condition for one of their patients.

In 2005, the American Academy of Pediatrics (AAP) endorsed a report from the American College of Medical Genetics (ACMG), which recommended that all states screen newborn infants for a core panel of 29 treatable congenital conditions and an additional 25 conditions that may be detected by screening (Appendix 1).[1] The Secretary of Health and Human Services' Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children (ACHDGDNC)† also adopted that report. Some states are now screening for more than 50 congenital conditions, many of which are rare and unfamiliar to pediatricians and other

*A note about language: although physicians often think of screening results as being "normal/abnormal" or "negative/positive," laboratories use the more specific language of "in range" and "out of range" to report results. We felt that it was appropriate to use and promote this language for the sake of clarity and consistency. For ease of reading, we use "parent" as a generic term to connote the adult who is responsible for a child's health care; we recognize that adults other than the biological parent may serve in this role. Where implications of congenital disorders are discussed, these obviously affect only those persons who are related biologically. In some circumstances, a primary care physician may suggest that the biological parent be contacted regarding congenital conditions, even if that parent is not the current primary caregiver for the child.

†A federal advisory committee to the Secretary of Health and Human Services, the ACHDGDNC advises and guides the Secretary regarding the most appropriate application of universal newborn screening tests, technologies, policies, guidelines, and programs for effectively reducing morbidity and mortality in newborns and children having or being at risk for heritable disorders.

primary health care professionals. In the foreseeable future, screening programs will likely adopt screening technologies that will further expand the number of conditions screened and tests offered.

The ACMG, with the support of the Health Resources and Services Administration (HRSA) Maternal and Child Health Bureau (MCHB), has developed and maintains Web-based resources it calls action (ACT) sheets to guide pediatricians through preliminary responses to out-of-range newborn screening results. These brief reference resources provide a focused, single-page summary of differential diagnoses, descriptions of the condition, actions to be taken by the pediatrician, diagnostic evaluation, clinical considerations, reporting requirements, and links to additional resources. ACT sheets are designed to be supplemented by state-specific information regarding referral resources. Many state-program Web sites have additional program-specific educational information; links to these program Web sites are readily accessible through an interactive map maintained by the National Newborn Screening and Genetics Resource Center (http://genes-r-us.uthscsa.edu/resources/consumer/statemap.htm).

Advances in newborn screening technologies and the availability of resources such as ACT sheets are aimed at improving health outcomes for affected children. To optimize this potential, primary care pediatricians (PCPs) must effectively engage the newborn screening program in their state. PCPs who treat patients who routinely cross state borders for care will likely engage multiple newborn screening programs.

The primary goals of this statement are to:

- delineate the responsibilities of PCPs and pediatric medical subspecialists within the newborn screening program;
- introduce 2 algorithms that, together, outline a clear and efficient pathway through the process of fulfilling those responsibilities; and
- outline resources that will support PCPs in addressing these responsibilities.

In addition to these primary goals, this statement addresses the steps that individual PCPs and practices must take to prepare for these responsibilities. We also recognize the significant roles other health care professionals and agencies have on the newborn screening system and identify ways these other entities can support PCPs and improve newborn screening and, therefore, advance improved health outcomes for newborns across the nation.

Limitations of This Statement

State newborn screening systems vary in their specific structure, procedures, and practices; this statement is focused on the core elements that are common to most state newborn screening systems. Newborn screening is increasingly being offered by commercial laboratories that market directly to parents and pediatric health care professionals. These programs introduce another layer of variation, which is beyond the scope of this statement.

Adequate funding of all aspects of newborn screening systems is necessary to ensure optimal performance of the system. This statement includes some general recommendations to promote such funding, and the AAP supports efforts to address financing for the nation's newborn screening systems and their constituent parts. Detailed recommendations for addressing the myriad challenges of system financing lie beyond the purview of this document.‡

Limitations of Newborn Screening

It is important to emphasize that newborn screening panels do not include all possible congenital conditions, and results for conditions on the panel should not be considered diagnostic. Thus, an in-range newborn screening result does not eliminate the possibility that a clinically symptomatic child has a congenital condition. Congenital conditions must be considered whenever an infant has signs or symptoms that are suggestive of (or consistent with) one of the disorders that can be detected by newborn screening.

An important goal of newborn screening is to identify infants with treatable congenital conditions before they become symptomatic. However, clinicians who care for children must be aware that some screened conditions may present with clinical deterioration before notification of newborn screening results. Pediatricians and emergency care physicians are often among the first health care professionals to encounter symptomatic infants, so they should be knowledgeable about the newborn screening program, ACT sheets for suspected conditions, and local or regional pediatric medical subspecialists to whom infants can be referred. The state newborn screening program usually can provide information about suspected conditions and expedite the newborn's follow-up confirmatory testing and care.

THE ALGORITHMS

The PCP plays several significant roles in the newborn screening system. In addition to responding to out-of-range newborn screening results, the PCP serves as a central source of education for parents regarding multiple aspects of the newborn screening system; the PCP also has responsibility for ensuring that newborn screening has been conducted, which can include providing education and encouragement to parents who decline screening. Finally, the PCP must ensure coordinated and comprehensive care for children affected by congenital conditions that are identified through newborn screening. The medical home provides a model for such care; the algorithms presented here address the specific roles of a medical home provider within the newborn screening system (Figs 1 and 2).

3- to 5-Day-Old Visit

The AAP[2] and *Bright Futures*[3] recommend neonatal follow-up visits in a child's medical home shortly after

‡For guidance on the *Current Procedural Terminology* (CPT) codes appropriate for use in the care of children who are identified as having congenital disorders, PCPs should refer to Rappo MA, Rappo PD. A special issue: coding for children with special health care needs. *AAP Pediatric Coding Newsletter*. January 2007. Available at: http://coding.aap.org/newsletterarchive.aspx.

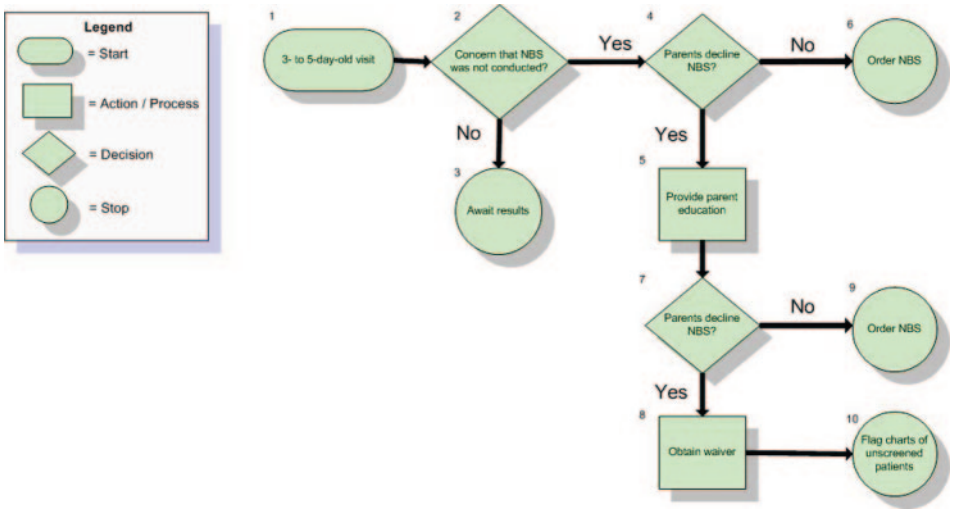


FIGURE 1
Algorithm 1. NBS indicates newborn screening program (see Appendix 2).

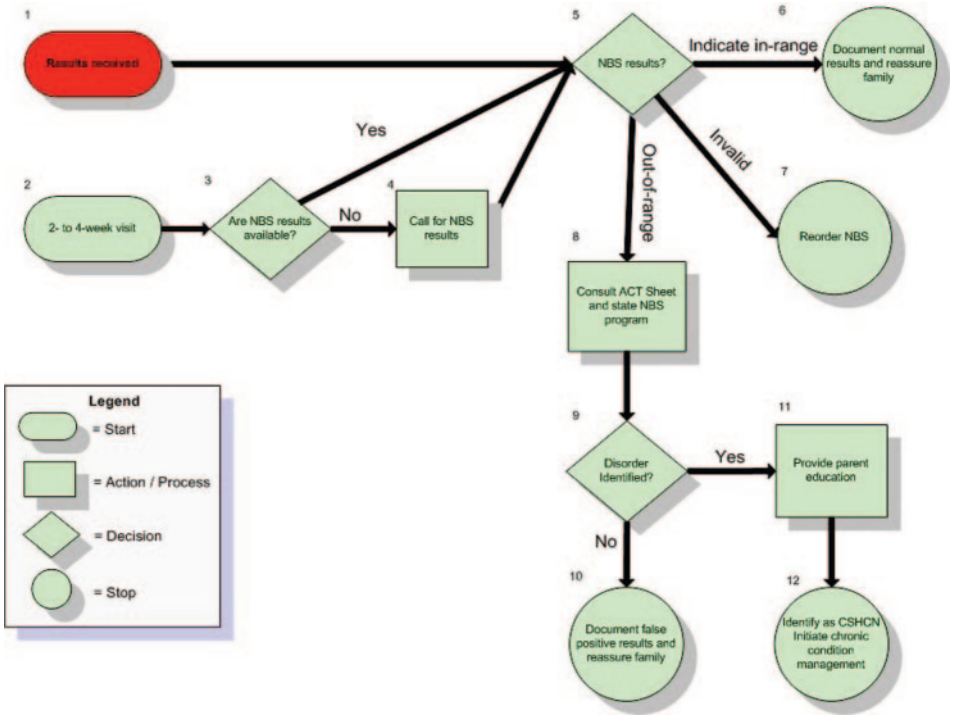
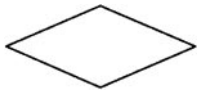


FIGURE 2
Algorithm 2. NBS indicates newborn screening program; CSHCN, child with special health care needs (see Appendix 3).

hospital discharge (3 to 5 days of life) and again by 1 month of age to ensure adequate weight gain, resolve neonatal concerns such as hyperbilirubinemia, and address parental questions. At the 3- to 5-day-old visit, the PCP should check for circumstances suggesting that newborn screening might not have been conducted.



Concern That Newborn Screening Was Not Conducted?

In most cases, newborn screening will occur as a result of standing orders at a hospital or birthing facility. In these cases, the PCP can address other aspects of the visit.

There are circumstances, however, under which the PCP might have cause for concern that the newborn screening was not performed. These circumstances include, but are not limited to, home births, emergency births, hospital transfers, and international adoption. In

addition, although most states mandate newborn screening, most jurisdictions provide parents with the right of refusal (see "Parents Decline Newborn Screening?").

If available discharge papers do not indicate that the newborn screening has been performed, the PCP should make arrangements for specimen acquisition.

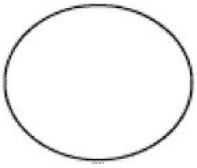
Parents Decline Newborn Screening?

If parental refusal is the reason that newborn screening has not been conducted, or if parents refuse newborn screening suggested by the PCP, the PCP should discuss the possible implications of nontesting and supply the parents with printed materials on newborn screening. Educational materials for parents and PCPs can be accessed through the AAP Web site (www.medicalhomeinfo.org/screening/newborn.html).

Provide Parent Education

Parent concerns and questions should be addressed fully, and a discussion of the general benefits and limited risks of newborn screening is recommended. More familiar conditions, such as congenital hearing loss, phenylketonuria, and sickle cell disease, may be used as examples.

If parental permission is obtained, arrangements for specimen acquisition should be made immediately, and newborn screening should be ordered.

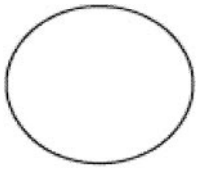


Order Newborn Screening

Newborn screening is conducted through the state newborn screening program, and protocols for ordering the screening vary by state. Contact information for each state's newborn screening program is available (see Appendix 4 and http://genes-r-us.uthscsa.edu/resources/consumer/statemap.htm).

Obtain Waiver

If parental permission is not obtained, parents or guardians should be asked to sign a waiver that documents their decision to decline newborn screening. In many cases, parents already will have signed a waiver at the hospital. PCPs should document the additional conversation and the parents' decision in the patient's chart and may wish to include a waiver signed in the PCP's office. A sample waiver form is included as Appendix 5; appropriate waiver forms should also be available through the state health department.



Flag the Charts of Unscreened Patients

In addition to documenting the discussion of newborn screening and the parents' refusal to consent to the screening, PCPs should flag the chart of any patients who are not screened so that the lack of screening will be taken into account should any subsequent concerns emerge regarding the child's growth or development. Vomiting, poor growth, seizures, developmental delay, lethargy, recurrent pneumonia, or poor feeding should prompt an evaluation that includes consideration of heritable conditions.

The chart note should also prompt the pediatrician to return to the question of newborn screening on subsequent visits to determine if the parents have changed their minds. The usefulness of screening after the neonatal period varies by condition, and use of state newborn screening systems for older infants varies by program.

Special Circumstances

For cases in which newborn screening is delayed because of previous parental refusal, because the infant was receiving total parenteral nutrition, or because of circumstances such as international adoption or an older infant entering care, the PCP should consult with the state newborn screening program regarding the availability and usefulness of the newborn screening protocol.

Newborn screening may not be ordered or may require an additional specimen in the case of preterm births, transfusion before screening, and other circumstances.[4] In these cases, the PCP should consult with a neonatal specialist.

In every circumstance, until and unless newborn screening is conducted, the patient's chart should be flagged to ensure that the lack of newborn screening is considered during ongoing care.

Results Received

In the case of an invalid or out-of-range screening result, the pediatrician identified on the newborn screening card should be called by the state newborn screening program in accordance with the urgency of the need for clinical intervention. In-range results are often transmitted by mail and should arrive before the 2- to 4-week visit.

2- to 4-Week Visit

The PCP cannot assume a "no news is good news" approach with regard to newborn screening. Delays or procedural failures at hospitals, state laboratories, other facilities, or within the newborn screening program may result in late or lost results. An infant's medical follow-up may not occur as planned, or newborn screening results may go directly to the child's birth facility instead of the infant's medical home.

Are Newborn Screening Results Available?

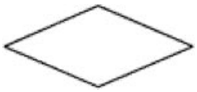
Office staff should check routinely for newborn screening results before the 2- to 4-week visit and pursue missing results before the visit. Using electronic-chart prompts or paper-chart templates for newborn visits will remind office staff to seek out newborn screening results.

Call for Newborn Screening Results

If newborn screening results are not available before the 2- to 4-week visit, the PCP should contact the state newborn screening program or the birthing facility for the results. An increasing number of state newborn screening programs have automated interactive telephone- or Internet-based systems through which pediatric offices can check for newborn screening results at any time.

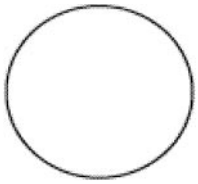
Special Circumstances

Occasionally, newborn screening results may be sent to the nonprimary physician; a physician who provides hospital or perinatal care for the infant may be noted on the newborn screening card even if he or she is not the infant's medical home physician. Clerical or other errors also may result in a physician who is unconnected to the child receiving the newborn screening results. However, the name on the card implies responsibility for the results, and physicians who receive results for patients who are no longer in their care should collaborate with the state newborn screening program and, in some instances, the hospital or birthing facility to locate the infant's family and/or current provider and to proceed with appropriate follow-up until the responsibility for subsequent care is clearly established. Physicians who receive results for patients with whom they or their colleagues have had no interaction should also notify the state newborn screening program immediately.



Screening Results?

The state newborn screening program will report results to the child's physician of record as being in range, invalid, or out of range. Appropriate responses to each of these results are discussed in the next sections.

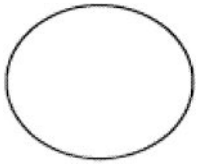


Document In-Range Screening Results and Reassure Family

In-range newborn screening results should be noted in the infant's chart and shared with the parents or guardians. In reassuring the family, the PCP should keep in mind that newborn screening does not rule out congenital conditions that are not included in the panel and does not absolutely guarantee the absence of the conditions that are screened. The PCP might note, however, that false in-range results are quite rare and the family can be reassured that their child is unlikely to be affected by conditions for which screening was performed.

Special Circumstances

Nine states (Arizona, Colorado, Delaware, Nevada, New Mexico, Oregon, Texas, Utah, and Wyoming) mandate an additional screening when the infant is 1 to 2 weeks old on the basis of the belief that a second screening is necessary to identify the maximum number of children with genetic disorders. A second screening is recommended for all infants in several other states, and approximately 25% of all US newborn infants currently receive 2 screenings. The relevance of second screenings for endocrinopathies is the subject of a study currently being designed by the MCHB. PCPs should familiarize themselves with their state's policies and procedures. If a second screening is ordered, it can be introduced and explained to parents within the context of state policies and the current limitations to newborn screening technologies discussed previously.



Reorder Newborn Screening

If the specimen is invalid (eg, collected too early, inadequate specimen, poor drying or application technique, inadequate or illegible patient information), the infant's newborn screening must be reordered and blood redrawn. This screen should be completed promptly to optimize the availability of results. PCPs must be familiar with local protocols for rescreening and should contact parents immediately to direct them to the site at which the second blood specimen will be obtained.

Consult ACT Sheets and State Newborn Screening Program

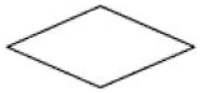
An out-of-range result on the newborn screening panel is not a diagnosis. However, some congenital conditions can be rapidly fatal in infants who appeared entirely healthy a few days earlier; thus, out-of-range screening results should always lead to prompt action by the PCP.

If the state newborn screening program does not provide the ACT sheet specific to the condition for which an out-of-range result was obtained, the PCP should download it (www.acmg.net/resources/policies/ACT/condition-analyte-links.htm).

The ACT sheet should be reviewed and followed in its entirety, but the most important actions are highlighted. These actions include:

- when to contact the family;
- whom to consult and whether an appointment is needed immediately;
- when the patient must be seen by the PCP;
- whether additional confirmatory testing is needed and what tests should be conducted;
- whether treatment is necessary and what treatment to initiate;
- how to educate parents about the condition; and
- when findings need to be reported back to the newborn screening program.

In addition to following ACT sheet recommendations, the PCP should consult with the state newborn screening program regarding out-of-range results. The state program should be familiar with local or regional experts for the conditions on their screening panels. In some states, the programs fund subspecialty clinics to conduct diagnostic evaluations and provide short-term and/or long-term subspecialty care to infants with out-of-range screening results.



Condition Identified?

After an out-of-range screening result is obtained, confirmatory testing and/or definitive consultation with subspecialists are required before a final diagnosis can be made.

To increase the sensitivity of a population screening test for rare conditions (and hopefully minimize the number of false in-range results missed), false out-of-range results are expected to occur, and false out-of-range results are significantly more frequent than true out-of-range results for most newborn screening tests. However, given the seriousness of the congenital conditions included in the newborn screening panel, the PCP must avoid complacency in the face of out-of-range results. Until confirmatory testing and/or definitive consultation with subspecialists can be accomplished, all out-of-range results must be taken very seriously.

Special Circumstances

In addition to true or false out-of-range results, confirmatory tests may identify the child as a carrier of the condition or may lead to an indeterminate result.

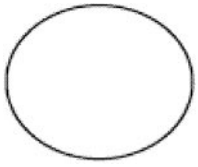
Carriers are individuals who are heterozygous for an autosomal-recessive condition and are usually not at risk of health problems themselves, although this may vary with the condition. Many state programs notify the PCP that the infant has been identified as a carrier, and it may be the responsibility of the PCP to disclose and discuss these results with the parents.

Knowledge of carrier status has 2 implications. First, because most of the conditions tested for on newborn screening are autosomal-recessive in inheritance, it is highly probable that at least 1 of the parents is a carrier also, and both parents might be carriers. If both parents are carriers, they have a 1-in-4 chance with each pregnancy of having an affected child. Alerting parents to the carrier status of their child serves to alert them that they may be at increased risk of having an affected infant with their next pregnancy. (When newborn screening results lead to genetic testing of the parents, pediatricians should be aware that misattributed paternity could be identified. Discussion with a geneticist or genetic counselor about how to manage these sensitive results may be helpful.)

The second implication of identifying a newborn as a carrier is that the infant will be at an increased risk of bearing an affected child when he or she achieves reproductive age if his or her future partner also is a carrier for the same condition. The risk is largely determined by the prevalence of the condition within the population, and additional genetic counseling may be warranted.

Occasionally, confirmatory diagnostic test results will not result in a definitive diagnosis. Uncertain results can be distressing to parents and PCPs, so thorough consultation with a subspecialist is essential. Unfortunately, indeterminate results may not be possible to resolve without more knowledge about some of these conditions and longer-term follow-up of these children.

At this point, it is incumbent on the PCP and the subspecialist to maintain an ongoing collaboration and continue to monitor the infant for signs and symptoms of a suspected condition. Children with uncertain results should have their chart identified for close monitoring. Good communication between the PCP and the consulting subspecialist is essential at this point to ensure that a unified message is conveyed to parents.



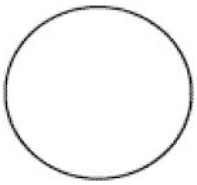
Document False Out-of-Range Results and Reassure Parents

In the event that the initial out-of-range result proves to be a false out-of-range result, the PCP can provide reas-

surance to parents. However, research that evaluated parents of infants with false out-of-range results has suggested that 5% to 20% of these parents will persist in their concerns about the health of their children for months or years after screening.[5–9] Therefore, PCPs should not take the event of a false out-of-range result too lightly and may wish to discuss this issue with parents on subsequent visits to provide additional reassurance and eliminate any misconceptions.

Provide Parent Education

To lay the foundation for comprehensive and collaborative care, it is critical during this time of uncertainty that the parents and family of the neonate be provided with condition-specific information and support as they await final clarification of the child's diagnosis and begin to plan for treatment and management. Parents are usually intensely anxious about the health of their child while the diagnosis is being pursued, and increasingly, parents are adept at tapping into resources on the Internet about specific conditions. Frequent, specific, and supportive communication from the PCP will help to avoid confusion and build trust. Appropriate materials for distribution to parents have been produced by the AAP (www.aap.org), the American College of Obstetricians and Gynecologists (ACOG [www.acog.org]), and MCHB/HRSA (www.mchb.hrsa.gov/screening). State newborn screening programs may also make educational materials available to health care professionals.



Identify the Child as a Child With Special Health Care Needs and Initiate Chronic Care Management

Any child who is given a diagnosis of a significant medical condition should be identified by the medical home physician as a child with special health care needs. Such a child should be entered into the practice's children with special health care needs registry, and chronic condition management should be initiated. Chronic care management provides proactive care for children with special health care needs, including condition-related office visits, written care plans, explicit comanagement with subspecialists, appropriate patient education, and effective information systems for monitoring and tracking the child's condition.[10]

IMPLEMENTING THE ALGORITHM

Role of the Medical Home

Regardless of diagnosis, every child needs a medical home to ensure coordinated and comprehensive care such that all of the medical, psychosocial, and educational needs of the child and family are met successfully within the local community. The PCP is responsible for providing a medical home.

Some conditions identified by newborn screening are relatively mild and or/transitory, and others have a wide spectrum of severity from asymptomatic to life-threatening crises. Plans for continuing care should be made in consultation with the family and appropriate subspecialists in light of the condition affecting the child and the severity of its manifestation. In some cases, the PCP may provide all or most of the ongoing care.

In other cases, the family may view their subspecialist as their primary care physician. Although a subspecialist may provide substantial ongoing care for a child who has been diagnosed with a severe and complex condition, the PCP retains the responsibility for providing a central source of "family centered, accessible, continuous, coordinated, comprehensive, compassionate, and culturally effective" care for the family.[11] The parents and family should be encouraged to maintain their relationship with their PCP. This relationship is critical, especially for cases in which the subspecialist is located at some distance from the family. In a crisis, the PCP may be the only available provider with knowledge of the child; he or she must have up-to-date information regarding the child's treatment.

The complex nature of many conditions identified by newborn screening may require care by a team of medical subspecialists, therapists, nutritionists, and educators.[12] The PCP and other professionals involved in the child's care must collaborate in the provision of acute care for illness or injury; surveillance of growth and development; anticipatory guidance to the family; immunizations; communications with schools, social services, and camps; transitions in care; and communication with other care professionals. In any case, clearly defined roles may help to reduce redundancies of services and prevent fragmentation of care.

The medical home should actively engage public and private resources to aid in the management of chronic conditions. Public health nursing provided through some state public health departments' maternal and child health programs often has a role in assisting PCPs, subspecialists, and families of children with conditions that are diagnosed through newborn screening. The level of public health nursing may vary from simply providing information and referrals to assisting with chronic condition management for a family.

If there is not a local health department or nursing service, PCPs may contact their state maternal and child health department (Title V) and the directors of programs for children with special health care needs through the state department of public health to obtain information on the availability of local family services; the state department of education for contacts with school nurses; and the early intervention agency (Individuals With Disabilities Education Act Part C) for contact information for the local early childhood connections program. Although the state resources for public health vary greatly from state to state, almost all communities have one or all of these resources available for

families. The national organization Family Voices (www.familyvoices.org) can provide information on local organizations and agencies that can offer resources to families with children with special health care needs and can assist families in accessing community services.

For additional information regarding care coordination, see the AAP policy statement "Care Coordination in the Medical Home: Integrating Health and Related Systems of Care for Children With Special Health Care Needs."[13]

Role of the Subspecialist

For most of the conditions that may be detected through newborn screening, the subspecialist will confirm the diagnosis, develop the treatment plan, educate the family about the treatment, monitor treatment, identify complications related to the disease process that may require additional referral, and work with other consultants in coordination of care. When acute illness exacerbates the condition, the PCP should work with the subspecialist to diagnose the acute illness and manage it appropriately to reduce morbidity.

Some children with conditions identified through newborn screening will have long-term sequelae that will require ongoing subspecialty management despite appropriate early intervention. Many of these children will have mild neurodevelopmental disabilities that may present as learning difficulties, attention-deficit/hyperactivity disorder, or other behavioral problems. However, in some instances, more significant cognitive and motor deficits and/or problems that adversely affect the child's feeding skills and respiratory status may be present. It is essential that the PCP provide ongoing screening and surveillance for these developmental disabilities.

Even with appropriate treatment, patients with certain conditions identified through newborn screening can undergo metabolic decompensation during an acute febrile illness. PCPs need to be aware of the initial clinical signs and laboratory abnormalities that may be found when metabolic decompensation occurs and be able to provide immediate intervention to stabilize the child until more specific advice can be obtained from the appropriate treating subspecialist. Effective communication among subspecialists as well as between each subspecialist and the PCP is essential for optimal long-term management of these children.[14] Long-term responsibilities of the subspecialist, in collaboration with the PCP, include:

- Providing genetic counseling and evaluation: Because the majority of conditions diagnosed through newborn screening are hereditary, genetic evaluation and counseling will be necessary for the parents. Older siblings may be affected with the condition but not yet symptomatic; diagnostic studies may be indicated for the siblings, and other relatives may wish to undergo carrier testing. The PCP, the state newborn screening program, and subspecialists are jointly responsible for ensuring that referral for genetic services occurs.
- Providing ongoing parent education and links to available resources: Resources for managing the condition should be made available to the patients and their families. Subspecialists, the PCP, and the state newborn screening program should collaborate in making appropriate referrals to programs for children with special health care needs, childhood early intervention programs, community-based support services, and additional subspecialists who are needed to evaluate and manage associated disabilities. Information from disease-specific advocacy organizations, along with parent brochures and guidance for child health care professionals, may be available through the subspecialist. The Genetic Alliance, a coalition of advocacy groups, serves as another national resource for parents (www.geneticalliance.org). The National Library of Medicine also has material on every condition in the expanded ACMG-recommended panel (http://ghr.nlm.nih.gov).
- Assisting in the transition to adult care: When transition to adult care is appropriate, the subspecialist will work with the PCP to identify a new team of physicians to care for the young adult. As adolescence proceeds, additional genetic counseling and preparation for family planning are appropriate.

RECOMMENDATIONS

Preparing the Practice

Before receiving notice of an out-of-range newborn screening result from their state newborn screening system, PCPs can take several steps to enhance their ability to successfully address their roles and responsibilities within the newborn screening program.

1. PCPs should familiarize themselves with their state newborn screening program via available (online) resources or, if necessary, by contacting the state program. PCPs should develop some familiarity with the conditions being screened and basic operations of their state newborn screening program, including protocols for retesting invalid screening results and conducting second screenings. PCPs should identify the person(s) with whom they should consult in the case of an out-of-range screening result and ensure that contact information is readily available.
2. State-specific contact information for regional pediatric medical subspecialists should be collected and kept on file in the PCP's office.
3. Procedures to address several steps of the algorithm should be developed in advance. These procedures include:
   a. updating contact information for the state newborn screening program and regional pediatric medical subspecialists;
   b. identifying children who are most likely not to have had newborn screening;
   c. confirming receipt of newborn screening results on all patients;

d. obtaining newborn screening results when they are not received from the state program;

e. documenting parental refusal of newborn screening; and

f. obtaining newborn screening specimens in the case of lost, delayed, or invalid results (the CPT code for retesting is 84030, and the diagnosis code is 270.10; PCPs should check with insurers to assess reimbursement).

4. PCPs should establish registries to identify, follow, and provide chronic condition management for children with special health care needs.

5. Educational materials regarding newborn screening should be on hand to distribute to expectant parents, parents who may decline newborn screening, and parents whose child's screening returns an out-of-range or inconclusive result. These materials should be available in languages and at literacy levels appropriate to all patients served. Appropriate materials for distribution to parents have been produced by the AAP (www.medicalhomeinfo.org/screening/newborn.html), ACOG (www.acog.org), and MCHB/HRSA (www.mchb.hrsa.gov/screening). State newborn screening programs may also make educational materials available to health care professionals.

Care coordination plays an essential role in ongoing efforts to integrate health and related systems of care for children and youth with special health care needs.[15] Becoming aware of available resources, being involved in the care coordination process, and developing unique care coordination approaches within one's own practice and community and in relationship with existing tertiary care centers are essential for providing optimal care for children with special health care needs. Families, PCPs, and other professionals can collaborate meaningfully to provide effective coordinated care.[13,15]

PCPs are also encouraged to participate in state, regional, or national registries; quality assurance programs; and/or research projects designed to enhance the care of children with the rare and complex conditions included in the newborn screening panel. They are also encouraged to seek opportunities for additional training and learning about state newborn screening programs and the conditions for which infants are screened and to work with their local AAP chapter and state newborn screening advisory committee (SNSAC) to advance the quality and effectiveness of the newborn screening system at the state and federal levels.

Collaboration With Other Health Care Professionals

The goals of ensuring the successful operation of the newborn screening system and advancing optimal care for infants and their families cannot be accomplished by PCPs alone. Effective collaboration and communication among PCPs and other clinicians and among the systems of care that engage the newborn screening system will ensure the best outcomes for infants and families. In light of this necessary collaboration, recommendations have been developed for prenatal health care professionals, hospitals and other birthing facilities, pediatric medical subspecialists, states and SNSACs, and federal agencies.

Prenatal Health Care Professionals

The prenatal period provides an ideal opportunity to begin to educate a family regarding the importance of newborn screening and the risks and benefits of early identification of the conditions identified through screening. The ACOG Committee on Genetics has asserted that "[o]bstetricians need to be aware of the status of newborn screening in their states and should be prepared to address questions or refer their patients to appropriate sources for additional information."[16] The following specific steps can help bring the awareness and knowledge of the obstetrician to bear in preparing a family for newborn screening and promoting the function of the newborn screening system.

1. Prenatal health care professionals are ideally positioned to educate expectant parents about the newborn screening program in conjunction with the prenatal screening program. The obstetrician is encouraged to begin the education early enough to allow patients the opportunity to ask questions that will assist them in understanding the purpose of newborn screening, its implementation, and the importance of test results and follow-up. Concise, clear, and comprehensive educational materials and/or video presentations already in existence should be made available to expectant parents during the prenatal period. Appropriate materials are available from the AAP (www.medicalhomeinfo.org/screening/newborn.html) and the National Library of Medicine (ghr.nlm.nih.gov/nbs).

2. Prenatal health care professionals should strongly encourage prospective parents to identify a medical home for their infant early in pregnancy. When the mother presents for postpartum care, the prenatal health care professional can further support the medical home by inquiring about the infant's well-being and follow-up care.

3. If an infant is lost to follow-up to the newborn screening program, prenatal health care professionals should assist in locating the family.

Hospitals and Other Birthing Facilities

In most cases, it is the facility at which the infant is delivered that is initially responsible for processing the newborn screening specimen. It is essential that these facilities have policies and procedures in place to ensure high-quality specimen processing and prompt delivery to the designated screening laboratory.

1. Particular attention should be brought to the development of protocols for:

   a. Repeat screening of invalid specimens.

   b. Documenting parental refusal to consent to newborn screening: Parents should be asked to sign a waiver form that documents not only their refusal to consent to newborn screening but also their

understanding of the program and its purpose and the risks associated with their refusal.

c. Adequate training of clinical and laboratory staff and quality assurance programs focused on high-quality specimen processing: Appropriate and complete information regarding the infant, contact information, and medical follow-up must be gathered and submitted with specimens.

d. Assisting public health authorities in locating infants who are lost to follow-up: If the infant's medical home is not clearly identified, the facility at which the child was born should assume responsibility for notifying the family of an out-of-range screening result and referring for additional diagnostic testing and subspecialty care.

2. Identification of the medical home or site of medical follow-up should be established as a condition for discharge.
3. Discharge materials should clearly indicate whether newborn screening was conducted and should identify the PCP and the in-hospital managing physician for later contact, if needed.
4. Hospitals and other birthing facilities should ensure the availability of printed and/or video educational materials, presented in concise and understandable language, to all families, including those whose primary language is not English. These materials should address the purpose of newborn screening, the risks and benefits associated with newborn screening, and the consequences of delaying or refusing newborn screening.
5. Opportunities for further discussion or questions should be made available with either the family's chosen PCP or staff members who are knowledgeable about the screening process and the conditions for which screening is conducted.

Pediatric Medical Subspecialists

Pediatric medical subspecialists play several roles in the care of children who have out-of-range results from newborn screening: they conduct confirmatory testing, care for the primary condition of infants who are affected by congenital diseases, and collaborate in the care of children with disabilities associated with some of the diseases identified through newborn screening. In fulfillment of these roles:

1. Pediatric medical subspecialists should assist the state newborn screening program in the development of educational materials for the public, families, PCPs, the state newborn screening program, and policy makers on specific conditions identified by newborn screening.
2. Pediatric medical subspecialists should serve on their SNSAC.
3. Pediatric medical subspecialists should respond promptly to requests for diagnostic and management services to infants with out-of-range screening results and children with conditions identified by newborn screening. Findings from clinic visits, laboratory studies, imaging studies, and diet and medication changes should be communicated promptly to the PCP, state newborn screening programs, other pediatric medical subspecialists, and the family (as appropriate).
4. Pediatric medical subspecialists should underscore the importance of maintaining a medical home relationship with the PCP for the infant identified with a condition through newborn screening.
5. Pediatric medical subspecialists should assist in the identification of associated disabilities and appropriate referral to other subspecialists for management.
6. Pediatric medical subspecialists should assist in the development of condition-specific protocols for the treatment of acute illness or injury and in the development of the child's care plan for school, activity restrictions, and special feeding/diet programs. Pediatric medical subspecialists should also work with the PCP, the family, and other subspecialists to delineate each person's role in managing acute illnesses, establishing relationships with schools and therapists, providing immunizations, working with social services and camps, and maintaining contact with insurers.
7. Pediatric medical subspecialists should provide ongoing education to the family and PCP about new developments and treatments for the condition and associated disabilities.
8. Pediatric medical subspecialists should work with the PCP and other subspecialists in identifying appropriate adult health care professionals for the transition to adult care.

State Systems

The state's role in newborn screening is to design, coordinate, and manage an effective newborn screening system. It has traditionally been the state's responsibility to oversee key aspects of the newborn screening system, including initial screening, confirmation of diagnosis, and coordination of short-term follow-up for infants with out-of-range screening results as well as longer-term care for children with special health care needs. Ultimately, the state must maintain an adequate public health infrastructure to ensure that every newborn infant receives appropriate care.

The AAP Newborn Screening Task Force set forth a broad agenda for state newborn screening systems in its statement published in 2000.[17] In addition to addressing the recommendations that follow, states are urged to consult that AAP statement for guidance in developing and supporting an effective and comprehensive newborn screening system.

To ensure the appropriate and effective function of newborn screening systems, the following recommendations must be addressed immediately:

1. States must monitor specimen collection and transmission of information between screening hospitals, the testing laboratory, and individual practitioners.

2. Identification of the follow-up medical home must be required on all newborn screening specimens.[16,18]
3. Laboratory collection and handling procedures must be clearly delineated at every site at which newborn screens are obtained or processed. State newborn screening laboratories are expected to maintain up-to-date technology and procedures and be prepared to implement recommended changes in the newborn screening process.[11]
4. Practical mechanisms should be established for retesting infants whose newborn screening results are indeterminate/invalid regardless of the cause.
5. Procedures should be adopted to ensure that the medical home is notified of out-of-range screening results by telephone on a schedule consistent with the urgency of the need for intervention. In the case of urgent out-of-range results, a designated medical subspecialist may be notified in addition to the medical home; the newborn screening program may need to contact the family if efforts to contact physicians are not successful.
6. Procedures should be adopted to ensure that in-range and invalid screening results are available to the medical home within 2 weeks of an infant's birth.
7. When out-of-range screen results are reported, the appropriate updated ACT sheet (or equivalent) and state-specific referral information should be forwarded immediately to the PCP.
8. States must have policies and procedures in place to locate children who have not established a medical home and to ensure that all newborn infants with out-of-range screening results receive appropriate diagnostic follow-up and subspecialty care.
9. States must provide clinicians with contact information for their newborn screening program coordinator and ensure that clinicians are updated promptly should any changes occur.
10. Public health agencies and maternal and child health programs should assist with care coordination for patients with special health care needs and their families.

Because states play a significant educational role in the newborn screening system, the following are recommended:

11. With direction from the SNSAC, states should develop and facilitate distribution of clear and concise educational materials for families at prenatal visits and in the hospital at the time of delivery. Condition-specific materials must be developed for families whose infants have out-of-range screening results; these materials include an explanation of test results, appropriate educational materials on the tested condition, referral for additional diagnostic testing, and referral for subspecialty care. Educational materials developed by the AAP, ACOG, and HRSA/MCHB may be used and/or supplemented with materials developed by the state. These materials can be accessed at www.medicalhomeinfo.org/screening/newborn.html or mchb.hrsa.gov/screening.
12. The state must develop educational information for medical professionals that outlines their responsibilities in the newborn screening process.

Finally, there are a number of steps that can be taken to improve the operation of the newborn screening system, including the following:

13. To prevent delays in processing when screening occurs on the weekend, the newborn screening laboratory responsible for state screening should operate at least 6 days a week, with coverage for holidays. Rapid turnaround time for results is essential for prompt diagnosis and treatment of metabolic conditions.
14. Information systems through which clinicians could directly download newborn screening results should be developed. Policies and regulations must be developed concurrently to protect privacy and confidentiality rights.
15. States should develop and implement information systems that facilitate the tracking of infants across state lines through communication and integration of data across newborn screening systems.
16. States must develop and implement policies that allow for interstate licensure and practice of medicine (including the use of telemedicine) to facilitate consultation and communication to underserved areas and ensure the free flow of information across state lines. There is a shortage of pediatric medical subspecialists across the country and a complete absence from more sparsely populated regions. This challenge must be addressed cooperatively by the states.
17. States should ensure the availability of ongoing care for infants with out-of-range screening results who lack health insurance and for those whose insurance does not provide coverage for necessary services and treatments. Medically required diets and vitamins are among the treatments often excluded from coverage provided by third-party payers.[19]
18. To promote greater understanding of the effects and benefits of the newborn screening system, states should develop information systems that are capable of tracking the multitude of performance measures for the newborn screening system and long-term outcomes of children with special health care needs identified through newborn screening. Performance measures include diagnosis for and treatment of infants with out-of-range screening results, cases missed by newborn screening, false out-of-range result rates, time to diagnosis, parental involvement and satisfaction, the social and psychological effects on families of infants with out-of-range and false out-of-range results, and family access to appropri-

ate and necessary services. Data to support the analysis of cost-effectiveness and cost benefit should also be collected.

19. To provide national data for newborn screening system quality assurance and program comparison, state programs should contribute timely case findings and laboratory data to the national newborn screening data-collection system operated by the National Newborn Screening and Genetics Resource Center (www2.uthscsa.edu/nnsis).
20. SNSACs should be authorized in each state to help implement and ensure the establishment of principles of universal access, clinician and community education, remedial surveillance for accountability, and quality of services for all infants. SNSACs should be chartered with appropriate authority and provided adequate support to effectively fulfill the roles outlined as follows.

State Newborn Screening Advisory Committees

1. SNSACs should comprise a balanced, representative, and diverse membership. Representation by diverse families and societal leaders should be balanced by members of the health care community, including clinicians in practice, representatives of hospitals and professional organizations, and public health experts, including the laboratories and the state. A diverse clinician representation would include pediatricians, obstetricians, family physicians, and nurse and midwife practitioners. In addition, the panel must have access to expert medical subspecialists, health care researchers, and biostatisticians.
2. SNSACs should cooperate with the US Department of Health and Human Services ACHDGDNC and other federal agencies to promote consistency in newborn screening throughout the nation.
3. SNSACs must work to advance state support and development of the newborn screening system, with particular attention to:
   a. efforts to use health information technology to advance clinician and family access to information about newborn screening as well as screening and follow-up services;
   b. optimization and accurate interpretation of privacy laws;
   c. implementation of a systems approach based on the Institute of Medicine principles for patient-centered safety, effectiveness, efficiency, timeliness, and equity[20];
   d. efforts to provide unfettered access, through both print and electronic media, to understandable education materials for families with diverse reading and language abilities; and
   e. development and distribution of resources for PCPs.
4. SNSACs must address identified challenges of fragmented service delivery as well as geographic, cultural, social, and financing barriers across county and state lines.
5. SNSACs should promote a statewide report on newborn health status for identifiable conditions and a national newborn health report that provides data on incidence, outcome, and community participation.
6. SNSACs must develop a mechanism for receiving feedback from parents, medical home practitioners, and subspecialists on the appropriateness of including particular conditions in the newborn screening program. This feedback should then be transmitted to the ACHDGDNC.
7. Each SNSAC is encouraged to develop its own charter and seek statutory establishment and state support.

National Partnerships

Although states remain responsible for newborn screening systems, federal agencies and national organizations play a significant role in the newborn screening system and in supporting families of children with genetic conditions. Strengthening national partnerships between federal agencies and professional, nonprofit, and family organizations provides the opportunity for a coordinated effort to increase the services offered to children with genetic and congenital conditions in all stages of diagnosis, treatment, and follow-up. There are 4 critical points of partnership for these groups: collaboration, funding, oversight, and follow-up.

Collaboration

1. Health care professionals, nonprofit agencies, state and federal public health programs, and families should seek to build relationships with other groups that focus on the newborn screening system. Relationships can be fostered through partnering on national initiatives, inviting other perspectives to serve on project advisory committees, and establishing a systematic method of receiving feedback from families.
2. Research should be performed on all aspects of newborn screening systems, including parent and provider education, results management, laboratory quality, residual specimen storage and use, and, most importantly, efficacy of newborn screening for each proposed condition. A national research agenda for newborn screening should be outlined. Input from federal agencies, professional associations, nonprofit organizations, and family support organizations should be coordinated. Multistate or national collaborations are often necessary to recruit a sufficient number of affected infants to understand the clinical spectrum of the disease and to compare treatment strategies. Collaboration will be key in conducting this research.
3. National partnerships should be developed and coordinated to support state newborn screening sys-

tems and encourage coordination, effective collaboration, and decrease duplication.

Funding

4. Adequate third-party reimbursement, grant applications, nonprofit fundraising efforts, and other sources of funding for newborn screening programs should be pursued by those who seek to improve the newborn screening system. Funding for the components of the newborn screening system and long-term care of children with genetic conditions comes from a variety of sources including screening fees, federal programs, state programs, nonprofit fundraising, insurance companies, and others, and such funding is critical at all levels.
5. Because ongoing research in the areas of education, results management, laboratory quality, and identifying and treating genetic diseases is important as the world of newborn screening continues to expand, funding for the implementation of these research projects should be provided.
6. Because establishing and funding a 24-hour hotline for access to online state-specific newborn screening program contact information can be useful in supporting state newborn screening programs, physicians, and families, a dedicated newborn screening hotline should be considered as part of preparing for national emergencies, natural disasters, or other circumstances.
7. Funding should be provided for demonstration projects directed toward strengthening the communication process between pediatricians and the newborn screening program. These efforts can include the development of telemedicine, effective health information exchanges, and linked information systems to facilitate the communication process.
8. Because the increased level of services required to comanage and coordinate care for patients with special needs identified through newborn screening can pose a significant financial burden for the PCP and the subspecialist, appropriate CPT coding that is aimed at enhanced reimbursement for chronic condition management should be developed.

Oversight

9. ACHDGDNC policies and activities should promote and facilitate uniformity across newborn screening programs, promote coordination between state newborn screening programs, support public health infrastructure for these programs, monitor the quality of these programs, and coordinate and promote research efforts related to newborn screening.
10. The ACHDGDNC should promote federal interagency collaboration and federal agency collaboration with state public health newborn screening programs to encourage coordination and effective collaboration between federal and state agencies.
11. Family involvement in all levels of newborn screening and follow-up care is important and should be encouraged. Families can give feedback on services provided, make suggestions on improving systems of care, advocate for needed services, and support other families that are going through similar situations.

Follow-up

12. Appropriate treatment and chronic condition management for children with congenital conditions should be ensured. Federal agencies, state newborn screening programs, and others can collaborate to create a national definition for follow-up to newborn screening systems.
13. Because enrolling children onto long-term research studies can provide the opportunity to test new treatments and better understand the natural history of chronic conditions, federal agencies and national organizations should promote opportunities for such research and create materials to educate parents about research in general and specific opportunities to participate in research.

National Medical Specialty Organizations, Including the AAP

National medical specialty organizations and their state chapters can play specific roles in the continued development of the collaboration necessary to ensure optimal performance of the newborn screening system throughout the country.

1. They should maintain communication with and participation on the ACHDGDNC to provide information to their constituencies and communicate any concerns to the ACHDGDNC.
2. They should foster education regarding newborn screening and promote pediatric medical subspecialties that focus on metabolic diseases among medical students and residents.
3. They should promote the development and implementation of a Health Plan Employer Data and Information Set (HEDIS) measure on newborn screening.
4. They should comment on the appropriateness of adding new tests to the core screening panel, ensuring that any newborn screening provides clear benefit to all children screened and to their families. These comments should be presented to the ACHDGDNC for consideration and adoption.

CONCLUSIONS

Advances in newborn screening technology, coupled with recent advances in the diagnosis and treatment of rare but serious congenital conditions that affect newborn infants, provide increased opportunities for positively affecting the lives of children and their families. These advantages, however, also pose new challenges to PCPs, both educationally and in response to the management of affected infants.

To respond appropriately, PCPs require immediate

access to clinical and diagnostic information and guidance; ACT sheets from the ACMG are a valuable source of such guidance. PCPs, however, have a proactive role to play in supporting the performance of the newborn screening system and ensuring the successful completion of their responsibilities to the program. PCPs must develop office policies and procedures to ensure that newborn screening is conducted and that results are transmitted to them in a timely fashion. PCPs must also develop strategies to use should these systems fail.

The newborn screening system extends well beyond the PCP's office, and many other stakeholders are essential for ensuring that the system functions well and supporting PCPs in their role within the system. The system is challenged by error, lack of education or information on the part of families and health care professionals, and systemic challenges such as the national shortage of pediatric medical subspecialists and barriers inherent in state licensing requirements. Lack of universal health care coverage and limited funding for newborn screening programs present additional significant challenges.

State and federal entities, hospitals, prehospital health care professionals, pediatricians, and pediatric medical subspecialists should act collaboratively to address these challenges or reduce their effects on the newborn screening system. AAP chapters and individual pediatricians should work together with the AAP and SNSACs to promote actions and policies that will optimize the function of newborn screening systems and ensure that children and families receive the full benefit of them.

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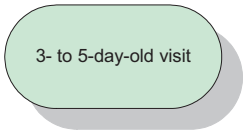
APPENDIX 1 2005 ACMG Recommended Screening Panel

OA	FAO	AA	Hemoglobinopathies	Other
Core panel				
Isovaleric acidemia	Medium-chain acyl-CoA dehydrogenase deficiency	Phenylketonuria	Sickle cell anemia (Hb SS disease) Hb	Congenital hypothyroidism
Glutaric acidemia type I	Very long-chain acyl-CoA dehydrogenase deficiency	Maple syrup disease	Hb S/β-thalassemia	Biotinidase deficiency
3-Hydroxy-3-methylglutaryl-CoA lyase deficiency	Long-chain L-3-hydroxy acyl-CoA dehydrogenase deficiency	Homocystinuria (caused by cystathionine β-synthase)	Hb S/C disease	Congenital adrenal hyperplasia (21-hydroxylase deficiency)
Multiple carboxylase deficiency	Trifunctional protein deficiency	Citrullinemia		Classical galactosemia
Methylmalonic acidemia (mutase deficiency)	Carnitine-uptake defect	Argininosuccinic acidemia		Hearing loss
3-Methylcrotonyl-CoA carboxylase deficiency		Tyrosinemia type I		Cystic fibrosis
Methylmalonic acidemia (Cbl A,B)				
Propionic acidemia				
β-ketothiolase deficiency				
Secondary targets				
Methylmalonic acidemia (Cbl C,D)	Short-chain acyl-CoA dehydrogenase deficiency	Benign hyperphenylalaninemia	Variant hemoglobinopathies (including Hb E)	Galactokinase deficiency
Malonic acidemia	Glutaric acidemia type II	Tyrosinemia type II		Galactose epimerase deficiency
Isobutyryl-CoA dehydrogenase deficiency	Medium/short-chain L-3-hydroxy acyl-CoA dehydrogenase deficiency	Defects of biopterin cofactor biosynthesis		
2-Methyl 3-hydroxy butyric aciduria	Medium-chain ketoacyl-CoA thiolase deficiency	Argininemia		
2-Methylbutyryl-CoA dehydrogenase deficiency	Carnitine palmitoyltransferase II deficiency	Tyrosinemia type III		
3-Methylglutaconic aciduria	Carnitine: acylcarnitine translocase deficiency	Defects of biopterin cofactor regeneration		
	Carnitine palmitoyltransferase I deficiency (liver)	Hypermethioninemia		
	Dienoyl-CoA reductase deficiency	Citrullinemia type II		

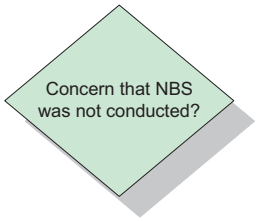
OA indicates disorders of organic acid metabolism; FAO, disorders of fatty acid metabolism; AA, disorders of amino acid metabolism; CoA, coenzyme A.

APPENDIX 2.

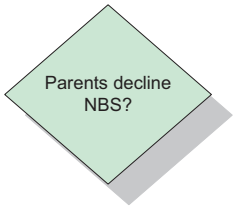
Algorithm 1



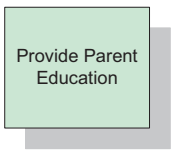
At the 3- to 5-day-old visit, the PCP should check for circumstances suggesting newborn screening might *not* have been conducted. Concerning situations include home or emergency births or international adoption.



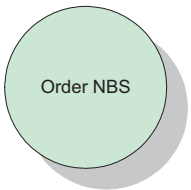
If the PCP has cause for concern that the newborn screening was not ordered and available discharge papers do not indicate the newborn screening has been ordered, the PCP should make arrangements for sample acquisition.



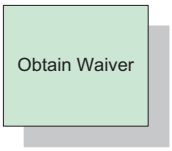
If newborn screening has not been conducted because of parental refusal or if parents refuse newborn screening suggested by the PCP, the PCP should discuss the general benefits and limited risks of newborn screening as well as possible implications of not testing.



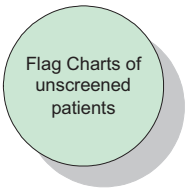
Educational materials for parents and PCPs can be accessed through the AAP Web site (www.medicalhomeinfo.org/screening/newborn.html). Parent concerns and questions should be addressed fully, and a discussion of the general benefits and limited risks of newborn screening is recommended. Very treatable conditions, such as phenylketonuria (PKU) and congenital hypothyroidism (CH), may be used as examples. If parental permission is obtained, arrangements for sample acquisition should be made immediately, and newborn screening should be ordered, as noted previously.



Newborn screening is conducted through the state newborn screening program, and protocols for ordering the screen vary by state. Contact information for each state's newborn screening program is available (http://genes-r-us.uthscsa.edu/resources/consumer/statemap.htm; Appendix 4).



If parental permission is not obtained, parents or guardians should be asked to sign a waiver documenting their choice to decline newborn screening. A sample waiver form is included as Appendix 5; appropriate waiver forms should also be available through the state health department.



PCPs should flag the charts of any patients who have not been screened so that the lack of screening will be taken into account should any subsequent concerns emerge regarding the child's growth or development. The chart note should also prompt the pediatrician to return to the question of newborn screening with subsequent visits to determine whether parents have changed their minds.

Special Circumstances

- In cases in which newborn screening is delayed, the PCP should consult with the state newborn screening program regarding the availability and usefulness of the newborn screening protocol.
- In the case of preterm births, neonatal transfusion, and other circumstances in which screening is not ordered or a second specimen is required, the PCP should consult with a neonatal specialist.

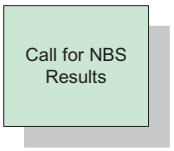
APPENDIX 3.

Algorithm 2

In the case of an out-of-range screening result, the pediatrician identified on the newborn screening card should be called by the state newborn screening program within the first week after birth. Normal results are often transmitted by mail; these may arrive before to the 2- to 4-week visit.

Delays or procedural failures at hospitals, state laboratories, or other facilities or within the newborn screening program may result in late or lost results. The PCP cannot assume a "no news is good news" approach with regard to newborn screening.

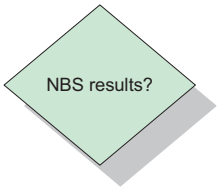
Office staff should check routinely for newborn screening results before the 2- to 4-week visit and pursue missing results in advance of that visit.



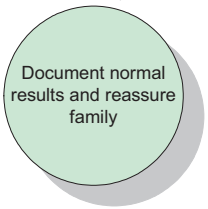
If newborn screening results are not available before the 2- to 4-week visit, the PCP should contact the state newborn screening program for the results at that time.

Special Circumstances

Newborn screening results may occasionally be sent to the wrong pediatrician. Physicians who receive results for patients no longer in their care should immediately contact the state newborn screening program and/or hospital to alert them that the PCP may not have received these results.



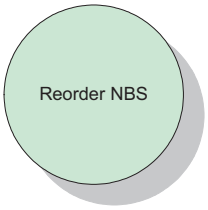
The state newborn screening program will report results as normal, unsatisfactory, or abnormal.



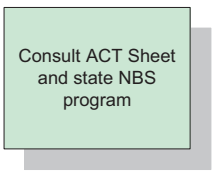
Normal newborn screening results should be noted in the infant's chart and shared with the parents or guardians. Keep in mind that newborn screening does not rule out congenital disorders that are not included in the panel and does not absolutely guarantee the absence of the disorders that are screened.

Special Circumstances

Currently, 9 states mandate a second screening, and 12 states allow it. If necessary, the PCP should order another newborn screening.



In instances where the specimen is unacceptable for testing, the infant's newborn screen must be reordered and a specimen must be obtained promptly.



If abnormal results are received, the PCP should access the ACT sheet specific to the disorder. The ACT sheet may be provided by the state newborn screening program or can be accessed at: www.acmg.net/resources/policies/ACT/condition-analyte-links.htm.

The most important actions to take are highlighted on the ACT sheets and include:

- When to contact the family;
- Whom to consult and whether an appointment is needed immediately;
- When the patient must be seen by the PCP;
- Whether further confirmatory testing is needed and what tests should be conducted;
- Whether treatment is necessary and what treatment to initiate;

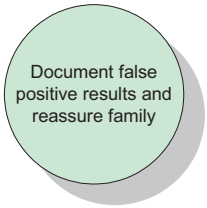
- How to educate parents about the disorder;
- When findings need to be reported back to the newborn screening program.

In addition to following ACT sheet recommendations, the PCP should consult with the state newborn screening program.

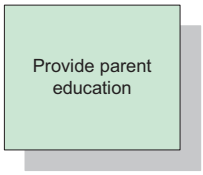
After an abnormal screening result is obtained, confirmatory testing and/or definitive consultation with subspecialists are required before a final diagnosis can be made.

Special Circumstances

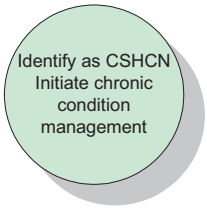
In addition to true or false positive results, confirmatory tests may identify the child as a carrier of the disorder or may be of indeterminate result.



In the event that the initial positive result proves to be a false positive result, the PCP can provide reassurance to parents. Up to 20% of these parents will persist in their concerns about the health of their children for months or years after screening.



To lay the foundation for comprehensive and collaborative care, it is critical during this time of uncertainty that the parents and family of the neonate be provided with disorder-specific information and support as they await final clarification of the child's diagnosis and begin to plan for treatment and management. Frequent, specific, and supportive communication from the PCP will help to avoid confusion and build trust.



The parents and family of the neonate should be provided with disorder-specific information and support as they await final clarification of the child's diagnosis and begin to plan for treatment and management.

Any child in whom a significant medical disorder is diagnosed should be entered into the practice's registry for children with special health care needs, and chronic care management should be initiated. Chronic care management provides proactive care for children with special health care needs, including condition-related office visits, written care plans, explicit comanagement with specialists, appropriate patient education, and effective information systems for monitoring and tracking the child's disorder.

A subspecialist may provide substantial ongoing care for a child with a severe and complex disorder. However, the PCP retains the responsibility to provide a medical home, which is the central source of "family-centered, accessible, continuous, coordinated, comprehensive, compassionate, and culturally effective" care for the family, and the family should be encouraged to maintain their relationship with their PCP.

The complex nature of many disorders identified by newborn screening may require care by a team of medical subspecialists, therapists, nutritionists, and educators. Clearly defined roles may help to reduce redundancies of services and prevent fragmentation of care.

APPENDIX 4 CONTACT INFORMATION FOR STATE NEWBORN SCREENING PROGRAMS

State	Contact	Web Site	Telephone	E-mail
Alabama	Melissa Tucker, AuD, CCC-A, Director of Newborn Screening Program	www.adph.org/newbornscreening	(334) 206–2944	melissatucker@adph.state.al.us
Alaska	Stephanie Birch, RNC, BSN, MPH, MCH Title V and CSHCN Director	www.hss.state.ak.us/dph/wcfh/screening_testing.htm	(907) 334–2424	stephanie_birch@health.state.ak.us
Arizona	Jan Kerrigan, RN, Program Manager	www.aznewborn.com/index.htm	(602) 364–1409	kerrigj@azdhs.gov
Arkansas	Dianne Pettit, Newborn Screening Coordinator	www.healthyarkansas.com/services/services_ph2.html#Newborn	(501) 280–4145	dpettit@arkansas.gov
California	John E. Sherwin, PhD, NBS Director	www.dhs.ca.gov/pcfh/gdb/html/nbs/	(510) 231–1728	jsherwin@dhs.ca.gov
Colorado	Vickie Thomson, MA, Director for the Newborn Screening Program	www.cdphe.state.co.us/ps/hcp/nbms/index.html	(303) 692–2458	vickie.thomson@colorado.edu
Connecticut	Vine Samuels, Newborn Screening Supervisor	www.dph.state.ct.us/BCH/NBS/NBS.htm	(860) 509–8081	vine.samuels@po.state.ct.us
Delaware	Betsy Voss, Newborn Screening Program Director	www.dhss.delaware.gov/dhss/dph/chca/dphnsp1.html	(302) 741–2987	betsy.voss@state.de.us
District of Columbia	Michelle Sermon, Genetics Program Specialist	http://doh.dc.gov/doh/cwp/view,a,3,q,573233, dohNav_GID,1802,dohNav,%7C33200%7C33245%7C.asp	(202) 442–9162	michelle.sermon@dc.gov
Florida	Lois Taylor, Director, Florida Newborn Screening Program	www.cms-kids.com/InfantScrning.htm	(850) 245–4670	lois_taylor@doh.state.fl.us
Georgia	Mary Ann Henson, MSN, Genetics Program Manager	http://health.state.ga.us/programs/nsmscd/index.asp	(404) 657–6359	mahenson@dhr.state.ga.us
Hawaii	Christine Matsumoto, Newborn Metabolic Screening Program Coordinator	www.state.hi.us/health/family-child-health/genetics/ genetics/nbshome.html	(808) 733–9069	chris.matsumoto@fhsd.health.state.hi.us
Idaho	Paige Fincher, Acting Manager	www.healthandwelfare.idaho.gov	(208) 334–4935	fincherp@idhw.state.id.us
Illinois	Claude-Alix Jacob, Deputy Director	www.idph.state.il.us/HealthWellness/genetics.htm	(217) 785–4093	claude.jacob@illinois.gov
Indiana	Iris Stone, Chief Nurse Consultant	www.in.gov/isdh/programs/nbs/NewbornScrSitemap.htm	(317) 233–1379	istone@isdh.in.gov
Iowa	Kimberly Noble Piper, State Genetics Coordinator	www.idph.state.ia.us/genetics/neonatal_parent_page.asp	(515) 281–6466	kpiper@idph.state.ia.us
Kansas	Melanie Warren, Follow-up Coordinator	www.kdheks.gov/newborn_screening	(785) 291–3363	mwarren@kdhe.state.ks.us
Kentucky	Sandy Fawbush, Nurse Administrator	http://chfs.ky.gov/dph/ach/ecd/newbornscreening.htm	(502) 564–3756, ext 3563	sandy.fawbush.ky.gov
Louisiana	Charles Myers, GSW, Program Administrator	www.dhh.louisiana.gov/offices/page.asp? ID = 263&Detail = 6302	(504) 219–4411	charlie@dhh.la.gov
Maine	John (Jack) A. Krueger, MSChE, Chief, Health and Environmental Testing Laboratory	www.maine.gov/dhhs/etl/newborn.htm	(207) 287–2727	john.a.krueger@maine.gov
Maryland	Susan Panning, NBS Director	www.fha.state.md.us/genetics/html/nbs_ndx.cfm	(410) 767–6730	panny.s.dhmh.state.md.us
Massachusetts	Roger Eaton, Director	www.umassmed.edu/nbs/index.aspx	(617) 983–6300	roger.eaton@umassmed.edu
Michigan	William Young, Newborn Screening Program Director	www.michigan.gov/mdch/0,1607,7–132–2942_4911_4916-64851—,00.html	(517) 335–8938	youngw@michigan.gov
Minnesota	Mark McCann, Supervisor, Newborn Screening Public Health Laboratory	www.health.state.mn.us/divs/fh/mcshn/nbshome.htm	(651) 201–5450; (651) 201–5471 (fax)	mark.mccann@state.mn.us
Mississippi	Beryl W. Polk, Director of Genetic Services	www.msdh.state.ms.us/msdhsite/index.cfm/41,0101,html	(601) 576–7619	bpolk@msdh.state.ms.us
Missouri	Sharmini V. Rogers, MD, BBS, MPH, Bureau Chief of Genetics and Healthy Childhood	www.dhss.mo.gov/NewbornScreening	(573) 751–6214	sharmini.rogers@dhss.mo.gov
Montana	Sib Clack, Newborn Screening Manager	www.dphhs.mt.gov/PHSD/family-health/newborn/newborn-screening.shtml	(406) 444–1216	sclack@mt.gov

APPENDIX 4 **Continued**

State	Contact	Web Site	Telephone	E-mail
Nebraska	Julie Miller, Program Manager	www.hhs.state.ne.us/nsp	(402) 471–6733	julie.miller@hhss.ne.gov
Nevada	[Vacant]	http://health.nv.gov/index.php		
New Hampshire	Marcia LaVochkin, Newborn Screening Program Coordinator	www.dhhs.state.nh.us/DHHS/MCH/default.htm	(603) 271–4225	mlavochkin@dhhs.state.nh.us
New Jersey	Mary Mickles, Program Manager of Newborn Screening and Genetics Services	www.state.nj.us/health/fhs/nbs/index.shtml	(609) 292–1582	mary.mickles@doh.state.nj.us
New Mexico	[Vacant]	http://sld.state.nm.us/nms	(505) 841–2581	
New York	Michele Caggana, MD, SCD FACNG Chief of Laboratory of Genetic Services	www.wadsworth.org/newborn	(518) 473–3854	mxc08@health.state.ny.us
North Carolina	Shu Chiang, Unit Supervisor Newborn Screening/Clinical Commissioner	http://slph.state.nc.us/Newborn/default.asp	(919) 733–3937	chu.chiang@ncmail.net
North Dakota	Barb Schweitzer, RN, Director of Newborn Screening	www.ndmch.com/metabolic-screening/default.asp	(701) 328–4538	bschweit@nd.gov
Ohio	William Becker, MD, Medical Director of Laboratory	www.odh.ohio.gov/odhPrograms/phl/newbrn/nbrn1.aspx	(888) 634–5227	william.becker@odh.ohio.gov
Oklahoma	Pam King, MPA, RN, Director of Genetics and State Genetics Coordinator	www.health.state.ok.us/program/gp/index.html	(405) 271–9444 ext 56737	pamk@health.ok.gov
Oregon	Cheryl Hermerath, Newborn Screening Program Manager	http://oregon.gov/DHS/ph/nbs/index.shtml	(503) 229–5882	cheryl.a.hermerath@state.or.us
Pennsylvania	M. Jeffery Shoemaker, PhD, Director	www.dsf.health.state.pa.us/health/cwp/view.asp?a=167&q=202513	(610) 280–3464	mshoemaker@state.pa.us
Puerto Rico	P. J.Santiago Borrero, Director of Hereditary Disease Program		(787) 754–3623	pjsantiago@centennial.net
Rhode Island	Ellen Amore, MS, Newborn Screening Program Manager	www.health.ri.gov/genetics/newborn.php	(401) 222–4601	ellena@doh.state.ri.us
South Carolina	Kathy Tomashitis, Pediatric Screening Follow-up Program, SC DHEC, Women and Children's Services	www.scdhec.net/health/lab/analyt/newborn.htm	(803) 898–0619	tomashk@dhec.sc.gov
South Dakota	Lucy Fossen, Newborn Screening Coordinator	www.state.sd.us/doh/NewbornScreening	(605) 773–2944	lucy.fossen@state.sd.us
Tennessee	Mitzi Lamberth, Newborn Screening Follow-up Program Director	http://health.state.tn.us/womenshealth/NBS/index.htm	(615) 262–6304	mitzi.lamberth@state.tn.us
Texas	David Martinez, Program Director	www.dshs.state.tx.us/newborn/default.shtm	(512) 458–7111, ext 2216	davidr.martinez@dshs.state.tx.us
Utah	Fay Keune, RN, Program Manager	http://health.utah.gov/newbornscreening	(801) 584–8256	fkeune@utah.gov
Vermont	Cynthia Ingham, Newborn Screening Program Chief	http://healthvermont.gov/regs/newborn_screening_reg.aspx	(802) 951–5180	cingham@vdh.state.vt.us
Virginia	Sharon K. Williams, RN, MS, Genetics Program Manager	www.vahealth.org/PSGS/	(804) 864–7712	sharonk.williams@vdh.virginia.gov
Washington	Mike Glass, Director	www.doh.wa.gov/ehsphl/phl/newborn/default.htm	(206) 418–5470	mike.glass@doh.wa.gov
West Virginia	Cathy Cummons, Newborn Screening Research Director	www.wvdhhr.org/mcfh/icah/unlinked/newborn_screening.htm	(304) 558–5388	cathycummons@wvdhhr.org
Wisconsin	Gary Hoffman, Newborn Screening Laboratory Manager	www.slh.wisc.edu/newborn	(608) 262–4692	hoffman@mail.slh.wis.edu
Wyoming	Shelly Gonzalez Sherry Long, Metabolic Records Analyst Nurse Consultant	http://wdh.state.wy.us/csh/index.asp	(307) 777–7943; (307) 777–7948	sgonza@state.wy.us; slong@state.wy.us

MCH indicates Maternal Child Health; CSHCN, children with special health care needs; NBS, newborn screening; SC DHEC, South Carolina Department of Health and Environmental Control.

THE AMERICAN ACADEMY OF PEDIATRICS STRONGLY RECOMMENDS NEWBORN SCREENING FOR ALL INFANTS

Refusal for Newborn Screening

Child's Name: _____ Child's Date of Birth: _____

Parent's/Legal Guardian's Name: _____

My child's doctor/nurse _____has advised me that my child (named above) should participate in the newborn screening program.

☐ As the parent or legal guardian of my child (named above), I choose to decline participation in my state's newborn screening program.

I have been provided information about newborn screening in my state and the importance of early identification of the diseases. I have had the opportunity to discuss these with my child's doctor or nurse, who has answered my questions regarding the recommended screening. I understand the following:

- The purpose of and the need for newborn screening.
- The risks and benefits of newborn screening.
- **If my child does not participate in newborn screening, the consequences of a late diagnosis of certain conditions can include mental retardation or death.**
- My child's doctor or nurse and the American Academy of Pediatrics strongly recommend that all newborn infants be screened for certain disorders.
- If my child has one of my state's screened conditions, failure to participate in newborn screening may endanger the health or life of my child.

Nevertheless, I have decided at this time to decline participation in the newborn screening program for my child, as indicated by checking the box above.

I acknowledge that I have read this document or it has been read to me in its entirety and I fully understand it.

Parent's/Legal Guardian's Signature _____ Date _____

Witness _____ Date _____

I have had the opportunity to re-discuss my decision not to participate in my state's newborn screening program and still decline the recommended participation.

Parent's initials _____ Date _____ Parent's initials _____ Date _____

Parent's initials _____ Date _____ Parent's initials _____ Date _____

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American Academy of Pediatrics
DEDICATED TO THE HEALTH OF ALL CHILDREN™

APPENDIX 5: DOCUMENTING REFUSAL TO HAVE INFANTS UNDERGO NEWBORN SCREENING

Despite the best efforts of health care professionals to educate parents and guardians about the need to have their infants undergo newborn screening and the importance of newborn screening in the early identification of certain diseases, some parents and guardians will decline to have their infants undergo newborn screening.

All parents and guardians should be informed about the purpose of and need for newborn screening, the risks and benefits of newborn screening, and the consequences of late diagnosis of certain conditions that would have been identified earlier through newborn screening. The use of this or a similar form that demonstrates the importance you place on newborn screening and focuses attention on the unnecessary risk for which a parent or guardian is accepting responsibility may, in some instances, induce a wavering parent or guardian to accept your recommendation.

Disclaimer. This form may be used as a template for such documentation, but it should not be used without obtaining legal advice from a qualified health care attorney about the use of the form in your practice. Moreover, completion of a form, in and of itself, never substitutes for good risk communication, nor would it provide absolute immunity from liability. For instances in which parents or guardians refuse newborn screening, health care professionals should take advantage of their ongoing relationship with the family and revisit the discussion on subsequent visits. Documentation in the medical chart of such follow-up discussions is strongly recommended, and the template, therefore, makes provision for this documentation.

This form may be duplicated or changed to suit your needs and your patients' needs and should be reviewed with your health care attorney before use. It will be available on the AAP Web[7] site (www.aap.org/bookstore).

Newborn Screening Expands: Recommendations for Pediatricians and Medical Homes Implications for the System

Newborn Screening Authoring Committee

Pediatrics 2008;121;192-217
DOI: 10.1542/peds.2007-3021