

# American Academy of Pediatrics Newborn Screening Task Force Recommendations: How Far Have We Come?

Michele A. Lloyd-Puryear, MD, PhD<sup>a</sup>, Thomas Tonniges, MD, FAAP<sup>b</sup>, Peter C. van Dyck, MD, MPH, FAAP<sup>a</sup>, Marie Y. Mann, MD, MPH, FAAP<sup>a</sup>, Amy Brin, MA<sup>b</sup>, Kay Johnson, MPH<sup>c</sup>, Merle McPherson, MD, FAAP<sup>a</sup>

<sup>a</sup>Health Resources and Services Administration, Rockville, Maryland; <sup>b</sup>American Academy of Pediatrics, Elk Grove Village, Illinois; <sup>c</sup>Johnson Consulting Group, Hinesburg, Vermont

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## ABSTRACT

The partnership of the Health Resources and Services Administration (HRSA)/Maternal and Child Health Bureau (MCHB) and the American Academy of Pediatrics (AAP) for improving health care for all children has long been recognized. In 1998, the establishment of the Newborn Screening Task Force marked a major initiative in addressing the needs of the newborn screening system. At the request of HRSA/MCHB, the AAP convened the task force to ensure that pediatric clinicians assumed a leadership role in examining the totality of the newborn screening system, including the necessary linkage to medical homes. The task force's report, published in 2000, outlined major recommendations for federal, state, and other national partners in addressing the identified barriers and needed enhancements of the care delivery system. Today, manifestations of the task force's recommendations are evident, many of which occurred under the leadership of HRSA/MCHB and the AAP. These activities are detailed in this article, with a discussion of future progression toward a quality, consistent, coordinated system of care for children identified with positive newborn screening results, their families, and the child health professionals who care for them.

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Dr Tonniges' current affiliation is Boystown Institute for Child Health Improvement, Omaha, NE; Ms Brin is currently enrolled in the School of Nursing, Vanderbilt University

### Key Words

newborn screening, medical home, system integration, federal initiatives, newborn screening task force, American Academy of Pediatrics

### Abbreviations

HRSA—Health Resources and Services Administration  
MCHB—Maternal and Child Health Bureau  
AAP—American Academy of Pediatrics  
NIH—National Institutes of Health  
CDC—Centers for Disease Control and Prevention  
AHRQ—Agency for Healthcare Research and Quality  
NNSGRC—National Newborn Screening and Genetics Resource Center  
ACMG—American College of Medical Genetics  
AAFP—American Academy of Family Physicians  
GPC—Genetics in Primary Care  
MS/MS—tandem mass spectrometry  
BOD—Board of Directors  
EHR—electronic health record  
PPI—Partnership for Policy Implementation  
MCH—maternal and child health  
EHDI—Early Hearing Detection and Intervention  
ACF—Annual Clinical Focus

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Address correspondence to Michele Lloyd-Puryear, MD, PhD, FAAP, Genetic Services Branch, Maternal and Child Health Bureau, Health Resources and Services Administration, 5600 Fishers Lane, Rockville, MD 20857. E-mail: [mpuryear@hrsa.gov](mailto:mpuryear@hrsa.gov)

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**I**N THE UNITED States, the early screening of children for special health care needs and congenital disorders begins in the newborn period for every infant in every state and every birthing center. Under the auspices of state public health agencies' newborn screening programs, all infants are tested universally for certain genetic conditions, such as hemoglobinopathies, metabolic disorders, hearing loss, and other congenital conditions. In addition, some states screen universally for infectious diseases such as HIV and toxoplasmosis.<sup>1</sup>

The advent of newborn screening began with the work of Asbjorn Folling in Norway almost 70 years ago, when he first defined what we now know as phenylketonuria. Public, state-based, newborn screening programs in this country began just over 40 years ago, with the development by Robert Guthrie of the filter paper-based testing technology still currently in use. Guthrie's innovation facilitated the development of states' newborn screening programs because the method was easy, stable, reproducible, inexpensive, and accurate, all necessary components of a universal, population-based, public health program. In the past 40 years, newborn screening programs have evolved from fragmented systems of public and private laboratory services, with disjointed follow-up monitoring, to the current state-based integrated systems. These programs are located within the state public health departments and operate through various partnerships or contractual relationships between public and private entities. Partnerships between the public health programs, pediatric clinicians, subspecialists, and childbearing families are essential to achieve fast effective diagnosis and treatment, and to prevent morbidity and death. Today, these newborn screening programs are faced with many challenges, including scientific advances in new screening technologies (and whether and how to introduce these new technologies), communication with the various partners essential in a well-functioning system, public concerns regarding privacy with respect to health information and the sharing of that information, and an ever-changing health care delivery system.

To understand more about the current status of newborn screening, a national task force was convened by the American Academy of Pediatrics (AAP) in 1998, at the request of the Health Resources and Services Administration (HRSA)/Maternal and Child Health Bureau (MCHB); it was funded by the HRSA/MCHB and National Institutes of Health (NIH) and was cosponsored by the Centers for Disease Control and Prevention (CDC), Agency for Healthcare Research and Quality (AHRQ), Association of State and Territorial Health Officials, Association of Maternal and Child Health Programs, Association of Public Health Laboratories, and Genetic Alliance.<sup>2</sup> The task force was organized to make additional recommendations for state newborn screening programs by using an approach that was based on the 6 compo-

nents of the newborn screening system, namely, screening, follow-up monitoring, diagnosis, treatment, evaluation, and education. More than 30 leaders, representing public health, medicine, and family perspectives, met over the course of 6 months, from autumn 1998 to summer 1999. At the end of the process, the task force put forth a national agenda for action and a series of recommendations to strengthen the newborn screening system, with proposed changes to meet the immediate technological and programmatic challenges facing these programs. These recommendations addressed (1) newborn screening public health infrastructure, (2) family and professional roles in the program, (3) oversight of newborn screening systems, (4) research and surveillance activities related to newborn screening, and (5) finance mechanisms supporting newborn screening systems.

During the appropriations process for fiscal year 1999 to 2000, there also was Congressional interest. Senate Finance Committee appropriations language indicated that

Within the funds provided, the Committee urges the availability and accessibility of newborn screening services to apply public health recommendations for expansion of effective strategies. HRSA, in collaboration with CDC and NIH, is encouraged to develop and implement a strategy for evaluating and expanding newborn screening programs, pilot demonstration projects, and use contemporary public health recommendations on specific conditions, such as cystic fibrosis and the fragile X syndrome. If implemented, the Committee directs that tangible steps be taken to protect patient privacy and to avert discrimination based on information derived from the screenings.<sup>3</sup>

Similar language was in the House appropriations report that fiscal year.

This article represents a review and analysis of the implementation of the task force recommendations by HRSA/MCHB and the AAP. They are the 2 primary organizations that either directly (through funding or provision of services) or indirectly (through guideline and policy development and AAP clinical guidelines) interact with state newborn screening programs. The recommendations relevant to these 2 organizations are presented in Tables 1 and 2.

## **ROLE OF THE MCHB AND TITLE V PROGRAMS IN IMPLEMENTATION OF THE TASK FORCE RECOMMENDATIONS**

### **History**

The history of Title V maternal and child health (MCH) programs and their genetic services activities are linked intimately to the history of newborn screening. HRSA/MCHB has responsibility for federal Title V activities and for administration of the current MCH services block grant supporting state activities. Federal funding for state-level programs and projects, through either block grant funding or discretionary funding, has been partic-

**TABLE 1** Activities Undertaken by HRSA

Recommendation	Action	Status
The federal government, acting through HRSA, CDC, HCFA (now CMS), AHRQ, NIH, and other agencies, should collaborate to provide ongoing leadership and support for development of newborn screening standards, guidelines, and policies.	Project with the ACMG (conditions and decision model)	Ongoing
HRSA/MCHB should strengthen current mechanisms to improve coordination of infant health programs and initiatives within the state and/or between states, including continuation of funding in support of newborn screening program reviews.	ACHDGDNC  NNSGRC established in 1999 Heritable Disorders Program established in 2004	Ongoing  Ongoing state reviews New cooperative agreements for regional collaboratives (7 regions and coordinating center)
A federally funded newborn screening research agenda should be outlined that aims to develop better tests (more sensitive, more specific, and less costly); to assess the validity and utility of new technologies (eg, MS/MS, DNA-based testing, and other evolving technologies); and to define appropriate uses of residual biological samples for population-based research and surveillance.	Undeveloped; current research projects include fragile X syndrome screening tool and hyperbilirubinemia screening nomogram	
HRSA/MCHB should provide grants to states to stimulate development of newborn screening information systems, with a focus on newborn screening systems that are connected to the medical home, newborn screening system process and outcome evaluation, development of standardized datasets, analyses of cost efficiency and cost-effectiveness, and integration with other public health data systems. Support for technological innovation (ie, new test technologies) should include these measures.	Integration of Child Health Information Systems Initiative	25 states funded with 38 grants (22 planning grants ended, 5 implementation grants ended in 2004, and 11 implementation grants are ongoing); PHII: development of policy/value case (ongoing), sourcebook/tool, and Community of Practice (ongoing); PHDSC: HRSA contract to develop recommendations regarding EHR/pediatrics (ongoing); AAP: EHR project (ongoing)
Pediatricians, pediatric subspecialists, and other health professionals who care for children should contribute to newborn screening data collection to advance knowledge about health outcomes and intervention effectiveness. Professional associations, the HRSA-funded NNSGRC, and state newborn screening programs should develop strategies to assist health professionals in their efforts to participate in and learn from newborn screening information systems.	Integration of Child Health Information Systems Initiative	PHDSC: HRSA contract to develop recommendations regarding EHR/pediatrics (ongoing); AAP: EHR project (ongoing)

ularly critical in establishing workable public health newborn screening systems within the states. Title V activities have been focused on (1) developing infrastructure for applied and clinical research, (2) fostering policy development, (3) ensuring service delivery, (4) improving program quality, and (5) providing education and training. Federal Title V funding and guidance have long been providing support for screening and diagnostic testing, follow-up services, nutritional supplements, and treatment services for children, as well as for state coordination efforts and public education. The manner in which MCH block grant funding to the states has been used for program support or implementation has been left to the discretion of the individual states.

The task force indicated that HRSA/MCHB should work with other public health agencies (NIH, CDC, and AHRQ), in partnership with public health organizations, child health professionals, and families, to implement a national agenda for strengthening newborn screening

programs. Because oversight of these programs is a state responsibility, the role of HRSA/MCHB has included Title V funding and technical assistance to states to foster policy development. HRSA/MCHB has focused its implementation efforts largely on addressing newborn screening public health infrastructure and family and professional roles in the newborn screening program. Fewer MCHB/HRSA activities were developed to address research activities. The bulk of federal support for basic research and implementation activities has been provided by the NIH, followed by the HRSA/MCHB, CDC, and AHRQ. For example, the NIH and HRSA/MCHB have been responsible for supporting program development, research for test development, policy development, and implementation of the state-based system of screening for such genetic conditions as phenylketonuria and sickle disease diseases. The CDC has been largely responsible for a system of quality assurance and proficiency testing through the National Quality Assurance

**TABLE 1 Continued**

Recommendation	Action	Status
<p>Federal and state public health agencies, in partnership with health professionals, families, and representatives of ethnic, minority, and other diverse communities, should develop model legislation and/or regulation that articulates policies and procedures regarding use of unlinked and identifiable residual samples for research and public health surveillance (this process should include review and consideration of the recent recommendations to the President set forth by the National Bioethics Advisory Commission for research involving human biological materials); develop model consent forms and informational materials for parental permission for retention and use of newborn screening samples; develop educational materials for parents that include information regarding the storage and uses of residual samples; organize collaborative efforts to develop minimal standards for storage and database technology to facilitate appropriate storage of residual newborn screening blood samples at the state level; consider creating a national or multistate population-based specimen resource for research in which consent is obtained from the individuals from whom the tissue is obtained; such a resource could be an alternative to retaining newborn screening samples for potential use in research.</p>	<p>Project with UCLA (educational process and residual blood specimens)</p>	<p>Discussion of use and storage of biological samples will be undertaken by ACHDGDNC; educational project through UCLA project: LSU toolkit for states for educational materials for parents (parent/provider focus groups in Maryland, Louisiana, New York, and Utah); NNSGRC review of policies for newborn screening, educating parents, genetics, residual blood spots, and informed consent</p>
<p>State MCH programs should conduct a review of the newborn screening system and its relationship to the HRSA/MCHB block grant performance measures and should evaluate the quality of data of the newborn screening-related performance measures.</p>	<p>Quality Assessment of the Newborn Screening System Initiative</p>	<p>Program evaluation and assessment scheme to develop a model based on existing guidelines that identifies quantifiable performance criteria for state newborn screening system components</p>
<p>States and the federal government should include public participation in medical policymaking. The Secretary's Advisory Committee on Genetic Testing (this advisory committee no longer exists and a new committee, the Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children, has been established. This committee will serve this mechanism) provides a mechanism for public participation in genetic policy development at the federal level. Each state should establish and fund a newborn screening advisory body with public participation to advise on newborn screening system policy developments. Such an entity should include a broad range of public advisors, representing parents, health professionals, third-party payers, appropriate government agencies, and other concerned citizens.</p>	<p>Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children</p>	<p>Ongoing</p>

ACHDGDNC indicates Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children; UCLA, University of California, Los Angeles; LSU, Louisiana State University; HCFA, Health Care Financing Agency; CMS, Centers for Medicare and Medicaid Services; PHII, Public Health Informatics Institute.

and Proficiency Program. This quality assurance program was cofunded by HRSA/MCHB for 25 years, until 2000.

### Improving Public Health Infrastructure

In the past 6 years, HRSA/MCHB responded to the task force recommendations by developing initiatives directed toward newborn screening public health infrastructure, including establishing a national center, the National Newborn Screening and Genetics Resource Center (NNSGRC); supporting state program improvements in program integration; implementing the Herita-

ble Disorders Program; promoting education for families and training for health care professionals; educating policymakers and newborn screening program staff members; conducting research; and developing guidelines and policy.

### NNSGRC

The mission of the NNSGRC is (1) to provide a forum for interactions among consumers, child health professionals, researchers, organizations, and policymakers, in refining and developing public health newborn screening and genetics programs, and (2) to serve as a national

**TABLE 2 From Task Force Report to Now**

Recommendation	Area of Effort	Title	Medium	Date/Status	Notes
The pediatrician should ensure that all newborns admitted to the practice have received adequate newborn screening and that appropriate documentation of testing is present.	Policy statements/technical reports/practice guidelines	Newborn Screening Fact Sheets	AAP Policy Statement by Committee on Genetics	Currently being revised	Multidisciplinary writing group organized
The pediatrician should assist the family in understanding the diagnosis, symptoms, and potential implications of a diagnosed genetic/metabolic condition, as well as the availability of genetic counseling, family testing, and other family support services.	Policy statements/technical reports/practice guidelines	Newborn Screening Fact Sheets	AAP Policy Statement by Committee on Genetics	Currently being revised	
The pediatrician should ensure that all newborns admitted to the practice have received adequate newborn screening and that appropriate documentation of testing is present.	Policy statements/technical reports/practice guidelines	Genetic Aspects of the Evaluation of the Child With Development Delay and Mental Retardation	AAP Policy Statement by Committee on Genetics	Currently being developed	
The pediatrician should ensure that all newborns admitted to the practice have received adequate newborn screening and that appropriate documentation of testing is present.	Policy statements/technical reports/practice guidelines	Technical Report: Congenital Adrenal Hyperplasia	Section on Endocrinology technical report	Reaffirmed in 2004	
The pediatrician should ensure that all newborns admitted to the practice have received adequate newborn screening and that appropriate documentation of testing is present.	Policy statements/technical reports/practice guidelines	Management of Hyperbilirubinemia in the Newborn 35 or More Weeks of Gestation	Subcommittee on Hyperbilirubinemia: Practice Guidelines	July 2004	
The pediatrician should ensure that all newborns admitted to the practice have received adequate newborn screening and that appropriate documentation of testing is present.	Policy statements/technical reports/practice guidelines	Health Supervision for Children With Turner Syndrome	AAP Policy Statement by Committee on Genetics	March 2003	
The pediatrician should ensure that all newborns admitted to the practice have received adequate newborn screening and that appropriate documentation of testing is present.	Policy statements/technical reports/practice guidelines	Health Supervision for Children With Sickle Cell Disease	AAP Policy Statement by Section on Hematology/Oncology	March 2002	

The pediatrician should ensure that all newborns admitted to the practice have received adequate newborn screening and that appropriate documentation of testing is present.	Policy statements/technical reports/ practice guidelines	Health Supervision for Children With Down Syndrome	AAP Policy Statement by Committee on Genetics	February 2001
The pediatrician should ensure that all newborns admitted to the practice have received adequate newborn screening and that appropriate documentation of testing is present.	Policy statements/technical reports/ practice guidelines	Maternal Phenylketonuria	AAP Policy Statement by Committee on Genetics	February 2001
The pediatrician should ensure that all newborns admitted to the practice have received adequate newborn screening and that appropriate documentation of testing is present.	Policy statements/technical reports/ practice guidelines	Health Supervision for Children With Williams Syndrome	AAP Policy Statement by Committee on Genetics	May 2001
The pediatrician should follow positive screening results to diagnosis, including repeated screening and diagnostic testing.	Policy statements/technical reports/ practice guidelines	Ethical Issues With Genetic Testing in Pediatrics	AAP Policy Statement by Committee on Bioethics	June 2001
The pediatrician should follow positive screening results to diagnosis, including repeated screening and diagnostic testing.	Policy statements/technical reports/ practice guidelines	Human Embryo Research	AAP Policy Statement by Committee on Bioethics and Pediatric Research	September 2001
The pediatrician should ensure that all newborns admitted to the practice have received adequate newborn screening and that appropriate documentation of testing is present.	Policy statements/technical reports/ practice guidelines	Evaluation of Newborns With Developmental Anomalies of External Genitalia	Committee on Genetics, Section on Endocrinology and Section on Urology	July 2000
The pediatrician should ensure that all newborns admitted to the practice have received adequate newborn screening and that appropriate documentation of testing is present.	Policy statements/technical reports/ practice guidelines	Molecular Genetic Testing in Pediatric Practice	AAP Policy Statement by Committee on Genetics	December 2000
The pediatrician should ensure that all newborns admitted to the practice have received adequate newborn screening and that appropriate documentation of testing is present.	Collaboration	Partnering with AAPF on genomics	AAPF's 2005 ACF incorporates the AAP Committee on Genetics representation	2004–2005

Genomics was identified as AAPF's 2005 ACF; AAP is partnering to ensure incorporation of pediatric perspectives, specifically regarding the areas of prenatal screening, newborn screening, and developmental disorders

**TABLE 2 Continued**

Recommendation	Area of Effort	Title	Medium	Date/Status	Notes
Parental knowledge should be reinforced after delivery through educational materials and discussion as needed by the infant's pediatrician or primary care health professional and/or knowledgeable hospital staff members.	Campaign	Safe First Week of Life Campaign	Campaign to address needs of mothers and practitioners to ensure optimal health during infant's first weeks of life	Ongoing	Campaign halted by lack of funds
Prospective parents should receive information about newborn screening during the prenatal period, preferably during a routine third-trimester prenatal care visit.	Campaign	Safe First Week of Life Campaign	Campaign to address needs of mothers and practitioners to ensure optimal health during infant's first weeks of life	Ongoing	Campaign halted by lack of funds
The pediatrician should maintain a central record and database containing all pertinent medical information about the child.	Expert meeting	EHR expert meeting	National meeting for experts in the EHR field to discuss integration of pediatric needs into development/implementation/functionality of EHR	September 2004	Recommendations from meeting will be presented to AAP BOD for consideration
The HRSA should engage in a national process involving government, professionals, and consumers to advance the recommendations of the task force and assist in the development and implementation of nationally recognized newborn screening system standards and policies.	Expert meeting/advisory	AAP representative appointed to Secretary's Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children	National advisory committee	2004 to ongoing	Stephen Edwards, MD, FAAP, appointed representative
The pediatrician should coordinate a seamless system of care with pediatric subspecialty clinics, tertiary care centers, and/or community-based providers when a child is diagnosed with a disorder through newborn screening.	State chapter activity	EHDI	AAP chapters through support of AAP State and Chapter Affairs Division and Division on Children With Special Needs	Ongoing	Integrated through role of "Chapter Champions"
The pediatrician should follow positive screening results to diagnosis, including repeated screening and diagnostic testing.	Expert meeting/advisory	Autism expert panel	AAP expert committee convened	2003 to ongoing	
The pediatrician should follow positive screening results to diagnosis, including repeated screening and diagnostic testing.	Expert meeting/advisory	AAP liaison appointed to attend the conference on Newborn Screening: State Policies and Procedures for Educating Parents about Newborn Screening and the Storage and Use of Newborn Screening Residual Blood Spots	HRSA/MCHB- and UCLA Center for Society, Individual, and Genetics- hosted conference	November 2001	Tracey Trotter, MD, FAAP, was appointed liaison

<p>The pediatrician should ensure that all newborns admitted to the practice have received adequate newborn screening and that appropriate documentation of testing is present.</p>	<p>State chapter activity</p>	<p>Chapters work to boost low screening rates</p>	<p>AAP chapter efforts</p>	<p>2000</p>	<p>Chapter efforts include education and advocacy regarding communication, documentation, and resources for increased use of EPSDT</p>
<p>Collaboration to provide ongoing leadership and support for development of newborn screening standards, guidelines, and policies.</p>	<p>Collaboration</p>	<p>Collaboration with ACMG to identify recommended screens</p>	<p>Steering committee for the AAP/ACMG project</p>	<p>2000</p>	
<p>The pediatrician should coordinate a seamless system of care with pediatric subspecialty clinics, tertiary care centers, and/or community-based providers when a child is diagnosed with a disorder through newborn screening.</p>	<p>Education/publication</p>	<p><i>Autism ALARM</i></p>	<p>Educational pamphlet on autism and primary care provider's role in diagnosis, management, and communication with family</p>	<p>January 2004</p>	
<p>Prospective parents should receive information about newborn screening during the prenatal period, preferably during a routine third-trimester prenatal care visit.</p>	<p>Education/publication</p>	<p><i>Caring for Your Baby and Young Child</i>, 4th ed</p>	<p>Softcover book</p>	<p>June 2004</p>	<p>Discusses guidelines for newborn screening</p>
<p>The pediatrician should coordinate a seamless system of care with pediatric subspecialty clinics, tertiary care centers, and/or community-based providers when a child is diagnosed with a disorder through newborn screening.</p>	<p>Education/publication</p>	<p>Childhood Hearing: A Sound Foundation in the Medical Home</p>	<p><i>Pedialink</i> module through AAP</p>	<p>July 2004</p>	<p>Online CME course addressing childhood hearing, including genetic causes of hearing loss</p>
<p>The pediatrician should coordinate a seamless system of care with pediatric subspecialty clinics, tertiary care centers, and/or community-based providers when a child is diagnosed with a disorder through newborn screening.</p>	<p>Education/publication</p>	<p>Surveillance and Screening in the Medical Home</p>	<p>Training curriculum</p>	<p>Spring 2003</p>	
<p>The pediatrician should assist the family in understanding the diagnosis, symptoms, and potential implications of a diagnosed genetic/metabolic condition, as well as the availability of genetic counseling, family testing, and other family support services.</p>	<p>Education/publication</p>	<p>Surveillance and Screening in the Medical Home</p>	<p>Training curriculum</p>	<p>Spring 2003</p>	



**TABLE 2 Continued**

Recommendation	Area of Effort	Title	Medium	Date/Status	Notes
The pediatrician should assist the family in understanding the diagnosis, symptoms, and potential implications of a diagnosed genetic/metabolic condition, as well as the availability of genetic counseling, family testing, and other family support services.	Education/publication	Interpreting Screening Tests to Families and Encouraging Follow-through	Article by Francis Glascoe, PhD	2002	
Prospective parents should receive information about newborn screening during the prenatal period, preferably during a routine third-trimester prenatal care visit.	Education/publication	Newborn Hearing Screening and Your Baby	Brochure	2002	Also available in Spanish
State public health agencies should direct their newborn screening programs to be consistent with professional guidelines and recommendations.	Training for physicians on how to advocate in their state for newborn screening to be consistent with AAP policy	AAP Chapter Advocacy Summit	Training seminar	November 2001	Highlighted advocacy strategies to enhance newborn screening
General education	General education for pediatricians on newborn screening	Contributing to AMA's white paper, Family Medical History in Disease Prevention	AAP Committee on Genetics will contribute to AMA's Family Medical History in Disease Prevention Article in <i>Pediatrics</i>	Ongoing	
General education	General education for pediatricians on newborn screening	What Follows Newborn Screening? An Evaluation of a Residential Education Program for Parents of Infants with Newly Diagnosed Cystic Fibrosis		August 2004	
General education	General education for pediatricians on newborn screening	Examination of the Communication Practices Between State Newborn Screening Programs and the Medical Home	Article in <i>Pediatrics</i>	February 2003	
General education	General education for pediatricians on newborn screening	Discovering Fragile X Syndrome: Family Experiences and Perceptions	Article in <i>Pediatrics</i>	February 2003	
General education	General education for pediatricians on newborn screening	Expanded Newborn Screening for Inborn Errors of Metabolism by Electrospray Ionization-Tandem Mass Spectrometry: Results, Outcomes, and Implications	Article in <i>Pediatrics</i>	June 2003	

General education	General education for pediatricians on newborn screening	Prospective Diagnosis of 2-Methylbutyryl-CoA Dehydrogenase Deficiency in the Himong Population by Newborn Screening Using Tandem Mass Spectrometry	Article in <i>Pediatrics</i>	July 2003
General education	General education for pediatricians on newborn screening	What's New in Newborn Screening?	NCE presentation sponsored by Committee on Genetics	October 2003
General education	General education for pediatricians on newborn screening	Newborn Screening by Tandem Mass Spectrometry for Medium-Chain Acyl-CoA Dehydrogenase Deficiency: A Cost-Effectiveness Analysis	Article in <i>Pediatrics</i>	November 2003
General education	General education for pediatricians on Newborn Screening	Newborn Screening Program Practices in the United States: Notification, Research, and Consent	Article in <i>Pediatrics</i>	February 2002
General education	General education for pediatricians on newborn screening	Diagnosis and Treatment of Maple Syrup Disease: A Study of 36 Patients	Article in <i>Pediatrics</i>	June 2002
General education	General education for pediatricians on newborn screening	Scientific Assessment of the Utility of Universal Newborn Hearing Screening	AAP Grand Rounds	June 2002
General education	General education for pediatricians on newborn screening	Universal Newborn Hearing Screening	Physician guide; AAP publication catalog	2002
General education	General education for pediatricians on newborn screening	Newborn Screening: Potentials, Challenges, and Politics	NCE presentation sponsored by Committee on Genetics	October 2002
General education	General education for pediatricians on newborn screening	<i>Guidelines for Perinatal Care</i> , 5th ed	Softcover book	November 2002
General education	General education for pediatricians on newborn screening	Genetic Counseling and Neonatal Screening for Cystic Fibrosis: An Assessment of the Communication Process	Article in <i>Pediatrics</i>	April 2001
General education	General education for pediatricians on newborn screening	Survey of Pediatrician Practices in Retrieving Statewide Authorized Newborn Screening Results	Article in <i>Pediatrics</i>	August 2001

CME indicates continuing medical education; AMA, American Medical Association; EPSDT, early and periodic screening diagnosis and treatment; NCE, National Conference and Exhibition.

resource center for information and education in the areas of newborn screening and genetics. Through its Web site, the NNSGRC provides newborn screening and genetic services information and resources to child health professionals, the public community, consumers, and state and federal government officials. It also assists states through technical assistance review; to date, >30 states have requested reviews for newborn screening and genetic programs. In addition to annual national data and information reports on state newborn screening activities, the NNSGRC has convened organizations and individuals for national discussions of pertinent topics in the areas of newborn screening and genetics. These discussions are to assist key decision-makers in the private and public sectors as they consider and respond to the challenges and opportunities that arise from scientific advances in genetics and newborn screening. Various smaller projects have been funded through the center, including development of a DNA-based screening mutation panel for cystic fibrosis for use in a diverse population; focus groups for the development of parent education materials on kernicterus; funding of a national hemoglobinopathy reference laboratory; and development of a manual on congenital adrenal hyperplasia for health care professionals.

The most recent activities of the NNSGRC, launched in 2004, include an additional cooperative agreement (ie, Quality Assessment of the Newborn Screening System) to work with state newborn screening programs and HRSA/MCHB to focus on the short- and long-term data needs of the screening programs (eg, screening and follow-up data). This project is evaluating previous program guidelines for newborn screening systems and will develop an evaluation and assessment tool that is based on those guidelines and identifies quantifiable performance criteria for state newborn screening system components.

### **Supporting State Program Improvements in Program Integration**

The task force also recommended specifically that HRSA/MCHB should use federal funding to “facilitate and foster the involvement of newborn screening systems in infrastructure development activities in states.”<sup>2</sup> Since 1999, HRSA/MCHB has provided financial and technical support to 25 state public health programs to promote the integration of newborn screening and related child health programs and their information systems, toward building a community child health profile.<sup>4</sup> The ultimate goal of these initiatives was to ensure that linkages to a medical home are made when a child is identified as screening positive in the newborn screening program. States’ projects focused on the development of standardized data sets for program and data integration, outcome evaluation, and analyses of cost efficiency and cost-effectiveness. With these funds, 16 states have integrated

child health information systems, linking their newborn screening programs with other child health information systems, including public, personal, and provider health information. HRSA/MCHB funding allowed the states to accelerate the process of integrating their information systems and provided the vision for integrating disparate information into a comprehensive, timely, and easily accessible record. HRSA/MCHB grantees reported that the idea of a “child health profile” was a catalyst in moving their departments toward integration. This was a new concept to them, and the value of having a community-based child health profile was immediately taken to heart. Missouri stated that the vision of a child health profile facilitated articulation of the importance of integration in meetings with stakeholders. For Indiana, the term “child health profile” helped provide a vision of something more than silo systems; it provided a vision of better communication with stakeholders. Washington State reported that the newborn screening team was able to host a retreat between stakeholders of the Early Hearing Detection and Intervention (EHDI) Program and the newborn dried blood spot screening program to build support for integrating the data collection processes and databases for these 2 public health programs. The successful retreat enabled the health department personnel to move forward rapidly with a combined data collection tool and to implement an integrated information system quickly. In Oregon, legislation was passed that required doctors to report to the EHDI Program. Legislators responded favorably because the EHDI Program was supported by HRSA and not state dollars. For Utah, receiving HRSA and CDC newborn dried blood spot screening grants at the same time allowed an integrated system to be organized. Without the grants, the dried blood spot screening program would have been behind schedule, and integration of the dried blood spot data would have been delayed.

Although HRSA/MCHB grant funding was small in comparison with the overall funds needed to build an integrated information system, this is evidence that a small amount of money from a federal agency can have far-reaching implications. The funding and the vision of a child health profile provided the impetus for states to articulate the need for an integrated child health information system in a manner that their stakeholders could understand easily. Having a federal agency willing to put money toward that vision carried significant weight with state health department policymakers and legislators.

A qualitative assessment of 7 state newborn screening programs’ integration efforts indicated that states needed tools to assist them in these integration efforts.<sup>5</sup> Toward that end, HRSA/MCHB coordinated the development of resource documents, *Integration of Newborn Screening and Genetic Services Systems With Other Maternal and Child Health Systems: A Sourcebook for Planning and Development*, and an assessment tool, *Integration of Newborn Screening*

*and Genetic Services Systems With Other Maternal and Child Health Systems: A Tool for Assessment and Planning.* These documents address the need for integration, present a logical description of essential functions that integrated information systems must support, outline a set of requirements to guide state public health programs in integrated information system development, and propose evaluation measurements. Newer initiatives include a cooperative agreement (ie, Newborn Screening Informatics Practice Network) with the Public Health Informatics Institute to assist state and local public health agencies in integrating child health information, ie, immunization records, newborn screening (hearing and dried blood spot) results, and vital statistics, by providing a peer-to-peer forum for the exchange of information about best practices in the development of integrated information systems.

### **Heritable Disorders Program**

In fiscal year 2004 appropriations language, Congress designated funds for the implementation of the Heritable Disorders Program. The Heritable Disorders Program was established to enhance, to improve, or to expand the ability of state and local public health agencies to provide screening, counseling, or health care services to newborns and children having or being at risk for heritable disorders. In response, HRSA/MCHB established a grant program to improve access to newborn screening and genetic services for medically underserved populations and to enhance activities such as screening and follow-up services, augmenting capacity needs, addressing needed training and education, improving subspecialty linkage, expanding long-term follow-up activities, strengthening linkages to medical homes, strengthening linkages to tertiary care, strengthening genetic counseling services, and enhancing communication/education for families and child health professionals. Through cooperative agreements, HRSA/MCHB funded 7 regional collaboratives across the country, linked together through a coordinating center. The American College of Medical Genetics (ACMG) was funded as the coordinating center for the collaboratives. The regional collaboratives represent the following regions and states: region 1: Connecticut, Massachusetts, Maine, New Hampshire, Rhode Island, and Vermont; region 2: District of Columbia, Maryland, New Jersey, New York, Pennsylvania, Virginia, and West Virginia; region 3: Alabama, Florida, Georgia, Louisiana, Mississippi, North Carolina, Puerto Rico, South Carolina, Tennessee, and Virgin Islands; region 4: Illinois, Indiana, Kentucky, Michigan, Minnesota, Ohio, and Wisconsin; region 5: Arkansas, Iowa, Kansas, Missouri, Nebraska, North Dakota, Oklahoma, and South Dakota; region 6: Arizona, Colorado, Montana, New Mexico, Texas, Utah, and Wyoming; region 7: Alaska, California, Hawaii, Idaho, Nevada, Oregon, Pacific Basin, and Washington.

The regional collaboratives have identified activities to address the disparities in services, including strengthening of regional telemedicine capacity, establishment of regional genetic service networks, and establishment of educational and training activities. The development of a network of genetic services and provider networks among the collaboratives will promote the identification of shared areas of need, as well as data collection and information sharing.

### **Promoting Education and Training for Families and Health Care Professionals**

Physicians and advanced practice nurses are dually charged to ensure appropriate care for newborns and to educate patients and their families properly regarding related health care issues. Because many parents are informed about newborn screening just before specimen collection, it is essential that all clinicians involved in the health supervision of newborns and expectant mothers be well informed about genetics and specifically about newborn screening. A survey by Kim et al<sup>5</sup> indicated that the techniques used most commonly to inform and to educate parents were informational brochures and a conversation with the parents (generally the mother of the newborn) immediately before or after the birth of the infant. The survey found, however, that states seldom defined who would have the role of and responsibility for informing and educating parents about newborn screening. There was also variability in the approaches and content of the state newborn screening educational materials. Often the family educational materials required a high school level of literacy to understand the content, which compromised the ability to ensure that parents had the understanding to make informed choices. Currently there is no national uniform policy for prescreening education or informing of parents. In addition, without national guidelines for content, there is no assurance that physicians and other health care professionals have the necessary tools to guide parents in the decision-making process. In 2003, HRSA/MCHB projects with Louisiana State University to develop educational materials for families and prenatal health care professionals were begun. These materials provide information about newborn screening for families and for health care professionals with the primary responsibility for prenatal health care and labor and delivery services (obstetricians, family practice physicians, and nurse midwives).<sup>6</sup> The materials were field-tested with the AAP, American Academy of Family Physicians (AAFP), and American College of Obstetricians and Gynecologists.

In September 1998, HRSA/MCHB launched the Genetics in Primary Care (GPC) project, with joint funding from the Bureau of Health Professions, HRSA, NIH, and AHRQ. This successful initiative was designed to plan, to implement, and to evaluate outcomes of faculty training

programs in genetics. The project focused its efforts specifically toward primary care (family medicine, general internal medicine, and general pediatrics) faculty members. Twenty medical schools and residency programs were funded through this project to train primary care faculty members to teach genetics. The outcome of this initiative was the development of additional specific newborn screening training and education projects for obstetricians, family physicians, and advanced-practice nurses.

Following up from the GPC project, the Society of Teachers of Family Medicine and the AAFP identified genomics as the topic for their Annual Clinical Focus (ACF) program for 2005. The ACF program is designed to develop educational materials based on the GPC curriculum and to bring awareness of the subject of genetics to AAFP members and the public. The ACF program for 2005 includes topics such as family history, newborn screening, developmental delay, counseling about genetic testing appropriateness and test results, and the role of genetic medicine in detecting and managing chronic diseases. HRSA/MCHB and NIH are collaborating to facilitate the development of specific modules on newborn screening, family history, and developmental delay for the ACF program.

#### **Educating State Policymakers and Newborn Screening Program Staff Members**

Ongoing education is important for all stakeholders involved in the newborn screening process. With that in mind, HRSA/MCHB developed genetics and newborn screening educational sessions for key state decision-makers, including state legislators and state health officials. These educational sessions were designed to create an environment for the participants to consider and to respond to the challenges and opportunities that arise from scientific advances in genetics and newborn screening, such as the represented states' genetic policies, programs, and activities; the legal framework for genetics and newborn screening for the represented states; the available services for children with special health care needs, particularly those identified through newborn screening; how state policies protect children from insurance discrimination on the basis of genetic or other health information; and the mechanisms in place to guarantee or to protect insurance benefits for children with genetic conditions.

After returning from the workshops, several attendants made legislative changes to strengthen or to address needs of their newborn screening programs (eg, Utah began screening universally for sickle cell diseases and Arizona enacted its hearing screening legislation). The need for educational sessions for senior policymakers continues to be felt as states face emerging fiscal and policy challenges.

An understanding by state program staff members of

the significance, functionality, and implications of using tandem mass spectrometry (MS/MS) is an important factor for forecasting future newborn screening needs. In 2001, HRSA/MCHB, recognizing the need for training in the use of MS/MS, established (in collaboration with the Association of Public Health Laboratories) training workshops for newborn screening program staff members, including laboratory personnel and personnel involved in newborn screening follow-up programs, to address the entire newborn screening system.

#### **Research**

Part of the task force national agenda was to "fund demonstration projects to evaluate technology, quality assurance, and health outcomes."<sup>2</sup> In 2001 and 2002, HRSA/MCHB funded 4 projects to evaluate various aspects of the introduction of new and evolving technologies into newborn screening programs, including the costs associated with the introduction of new technology; parent education; ethical, legal, and social issues; and the development of the technology itself.

In 2001, the Departments of Health of California and Massachusetts each were awarded a 3-year grant to identify models for evaluating the clinical validity, utility, and costs associated with the use of MS/MS in newborn screening programs. Massachusetts was partnered within its regional network with Maine, New Hampshire, Vermont, and Rhode Island to identify data elements (model database, data entry and analyses, screening algorithms, and case definitions) that would be useful in analyzing MS/MS and cystic fibrosis screening. California was partnered with Alaska, Hawaii, Washington, and Oregon on the project titled Implementation of New and Innovative Technologies into Newborn Screening Programs.<sup>7,8</sup>

In 2001, the Department of Health in Hawaii, in a multistate partnership with Alaska, California, Idaho, Washington, and Oregon, was funded to examine the cost of parental informed consent; parental attitudes toward informed consent; family-based development of newborn screening educational materials; provider and consumer educational materials; and parental attitudes toward the ethical, legal, and social implications of expanded screening.<sup>9</sup> The project reported that most women did not recall receiving any information about newborn screening. All of the participants wanted the information from their prenatal care provider before giving birth. In addition, most participants thought that, if they were informed about newborn screening prenatally, the current system of "informed dissent" was adequate. All women in the focus groups also approved of screening for disorders with no proven treatment that can be given in the newborn period. However, there was significant disagreement about screening newborns and children for adult-onset disorders.

In 2002, the Greenwood Center was funded to de-

velop a screening tool for fragile X syndrome that would be suitable for universal population-based screening. That project is ongoing.

In 2004, additional funds were targeted toward reducing the incidence of severe hyperbilirubinemia and kernicterus among term and near-term healthy newborns. That project is ongoing and will assess prospectively and validate published methods for predicting the incidence of hyperbilirubinemia during the first 2 weeks of life.

Although these completed projects were effective, a significant federally funded research agenda has yet to be implemented. Recommendations for a research agenda were put forth by the ACMG Newborn Screening Expert Group.<sup>10</sup>

### **Guidelines and Policy Development**

The task force recommended that HRSA/MCHB “engage in a national process involving government, professionals, and consumers to advance the recommendations of this Task Force and assist in the development and implementation of nationally recognized newborn screening standards and policies.”<sup>2</sup> The interpretation and implementation of guidelines vary considerably among state programs. Programmatic differences include the number of conditions screened for, criteria used to select which conditions are screened for, fee structure, laboratory capacity, and newborn screening follow-up system.

The task force’s recommendations reflected the need to develop national guidelines for newborn screening programs, to ensure equitable access to newborn screening. For this reason, HRSA/MCHB contracted with ACMG to analyze the scientific literature and to gather expert opinions, to examine the available information concerning the scope of newborn screening programs and anticipated future directions, including delineation of the best evidence for screening for specified conditions, and to develop a set of recommendations based on this best evidence that would address (1) a uniform condition panel (including implementation methods), (2) model policies and procedures for state newborn screening programs (with consideration of a national model), (3) minimal standards for state newborn screening programs (with consideration of national oversight), and (4) a model decision matrix for consideration of state newborn screening program expansion. The project was reported by the ACMG Newborn Screening Expert Group<sup>9</sup> and was presented to the Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children for review and recommendations.

Perhaps the most significant policy development initiative occurred with the establishment of the Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children in 2003. This committee advises the Secretary of the Department of Health and Human Services concerning the grants and projects au-

thorized under the legislation, as well as the most appropriate application of universal newborn screening tests, technologies, policies, guidelines, and programs for effectively reducing morbidity and mortality rates for newborns and children having or being at risk for heritable disorders. The committee is staffed by HRSA/MCHB, and agenda development is coordinated with other federal, ex officio members from CDC, NIH, and AHRQ. Information regarding the committee’s charter, its legislation, and its meetings may be found on the MCHB Web site (<http://mchb.hrsa.gov/programs/genetics/committee/default.htm>).

### **THE AAP: PROMOTING THE ROLE OF THE MEDICAL HOME IN NEWBORN SCREENING**

#### **Partnerships**

Since the 1999 publication of the task force report, the AAP has partnered with several national organizations to assist in enhancing the public health infrastructure and promoting research protocols aimed at newborn screening. These partnerships, as well as AAP efforts in education, standard setting, and resource development, are framed in the AAP belief that all children should have access to a medical home.<sup>11</sup> The AAP defines the medical home as the ideal place for a child to receive accessible, family-centered, continuous, coordinated, comprehensive, compassionate, and culturally effective care. A child who has a medical home has a primary child health professional who works in partnership with the child’s family to ensure that all medical, nonmedical, psychosocial, and educational needs of the child and family are met. This quality-based approach to care is an essential part in the newborn screening process.

#### **Education**

The positive identification of a congenital disorder through newborn screening presents a complex set of variables to child health professionals, such as the low incidence of many of the disorders, the complexity of the associated symptoms, and the emotional tension of the family with a positive screen. With these variables, the objectives of the educational efforts of the AAP are focused on enhancing the medical home provider’s ability to provide quality care to identified newborns and children and educating parents about the complex system and their role within it.

Provider education has involved several approaches, such as educational sessions at the annual AAP national conference and exhibition; articles in *Pediatrics* that discuss current technology, genetic counseling, and the current status of newborn screening program linkages with the medical home; and development of the 2003 *Surveillance and Screening in the Medical Home* training curriculum. Through its partnership with the CDC National Center on Birth Defects and Developmental Dis-

abilities, the AAP launched the *PediaLink* module “Childhood Hearing: A Sound Foundation in the Medical Home” in 2004 (available at [www.pedialink.org](http://www.pedialink.org)). This Web-based educational module educates child health professionals on different types of hearing loss, screening methods, medical and genetic risk factors, the importance of early detection, and the critical role of the medical home.

The AAP believes in the many benefits of educating parents, facilitating the construction of family-professional partnerships. This belief has been illustrated in past publications of *Newborn Hearing Screening and Your Infant* brochures, inclusion of newborn genetic/metabolic and hearing screening material in the fourth edition of the bestseller *Caring for Your Infant and Young Child*, and parental fact sheets on hyperbilirubinemia. In June 2005, the AAP Board of Directors (BOD) endorsed a series of new consumer and provider educational materials regarding newborn genetic/metabolic and hearing screening. Developed by health literacy researchers at Louisiana State University through a contract with MCHB, these materials confronted the literacy chasm contained in current state newborn screening materials.<sup>6</sup> Distributed to members of the AAP, National Pediatric Nurse Practitioner Association, American Academy of Physician Assistants, and March of Dimes chapters in early autumn of 2005, these materials offer providers new opportunities to communicate with their patient’s families about the newborn screening process, what to expect from it, and the significance of it.

### Standard Setting

Since the 1999 task force, the AAP continues to engage in discussions regarding clinical guidance for child health professionals on the health supervision of metabolic/genetic/infectious/hearing disorders. The AAP commentary on newborn screening dates back to the 1965 statement *Screening of Newborn Infants for Metabolic Disease*.<sup>12</sup> Current policy statements range from specific diseases to ethical implications of screening, while universally promoting child health professionals’ critical role throughout the process. For example, the Committee on Genetics newborn screening fact sheets provide a summary of the many issues involved in newborn screening, as well as a concise listing of pertinent information according to specific genetic diseases. The revised fact sheets were presented to the AAP BOD in 2005.

In the summer of 2005, the AAP established an expert, multidisciplinary, authoring body, from the pediatric domains of primary care, genetics, neonatology, neurodevelopment, quality improvement, and informatics, to draft an operationalized clinical report targeted at the point of care. The report will delineate explicitly primary care child health professionals’ role in quality care for identified newborns and children, specifically defining their role in comanaging the condition

with a subspecialist and the family. It is the hope of the AAP that this clinical report will facilitate identification of clinical standards of care for these newborns and children, with future opportunities to evaluate its implementation in practice. The AAP acknowledges that it is through ongoing evaluation of such standards that the pediatric community can ascertain best practice models.

Moreover, the AAP recognizes the tremendous significance the Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children has in shaping the future of newborn screening in the United States. The committee’s recommendations regarding the implementation of expanded newborn screening will affect child health professionals directly. The role of Stephen Edwards, MD, FAAP, past president of the AAP, on the committee as a representative from the medical community is critical in linking those recommendations to the development of future clinical standards. The AAP thinks the clinical guidance it provides should be reflective of the infrastructure established in states, practices, and communities.

### Resource Development

Through funding from HRSA/MCHB, the AAP National Center for Medical Home Initiatives, which is housed in the Division of Children With Special Needs, has developed a variety of resources to enhance the medical home’s provision of care for newborns and children with rare disorders. In 2003, the center launched an outreach effort focused on the inequity in states’ sickle cell disease screening practices. Aiming at the 7 states that were not screening for sickle cell diseases at that time, targeted outreach activities, site visits, and educational efforts were conducted. These activities helped lead to universal implementation of sickle cell disease screening (New Hampshire screens for sickle cell disease only in selected populations or by request).<sup>1</sup> Accompanying this outreach effort was the development of an educational sickle cell disease toolkit. These toolkits were distributed to all AAP members and to all AAP and AAFP chapters in the summer of 2004.

Before the middle 1990s, the average age of identification of hearing loss in the United States was 30 months.<sup>13</sup> To engage in this cause, the National Center and the Department of State and Chapter Affairs established a national network of professionals in the AAP state chapters to promote hearing screening and its necessary linkage to a child’s medical home. These “Chapter Champions” have been instrumental in educating child health care professionals, at the state and local levels, on necessary identification, diagnosis, treatment, and follow-up guidelines developed by the AAP Task Force on Improving the Effectiveness of Newborn Hearing Screening, Diagnosis, and Intervention. These guidelines, titled *Universal Newborn Hearing Screening Diagnosis and Intervention Guidelines for Pediatric Medical Home Pro-*

viders, are presented in an algorithm format to assist child health professionals in the treatment and follow-up evaluation of a child diagnosed with a hearing loss. The algorithm provides visual cues regarding all necessary points of referral and communication for comprehensive coordinated care.

Many infants who have conditions detected through newborn screening soon develop medical conditions and disabilities that could qualify for and benefit from state early intervention programs (operated under part C of the Individuals with Disabilities Education Act). Because of the significant relationship between medical homes and early intervention programs, the HRSA/MCHB, the AAP National Center, and the Office of Special Education Programs in the Department of Education conducted a periodic survey in 2002 to assess pediatricians' perceptions of, involvement with, and barriers to referral for early intervention. Results from that survey highlighted a mutual gap in awareness between the medical home and early intervention professionals. To increase familiarity, a brochure that outlines the roles of the medical home and early intervention professionals in coordinating care for an identified child, provides sample forms to assist in communication, and lists additional resources for professionals and family members was developed. The brochure was distributed to all AAP members in February 2005 and was given to all state part C coordinators.

Another barrier identified in the 2002 periodic survey was the nonuniversal referral process for states' early intervention service programs. The referral process itself, as well as the patient information needed to make a referral, varied from state to state. In an attempt to offer a universal referral form containing only the consistently required patient information, the AAP National Center, the Office of Special Education Programs, and the Orelena Hawks Puckett Institute hosted a meeting with primary pediatric care and early intervention professionals in December 2004. Participants drafted a universal referral form, which was test piloted throughout the nation in the summer of 2005. Subsequent plans to pilot test the form through appropriate AAP channels before submission to the BOD in 2006 are currently underway. All of the materials described in this section can be downloaded from the National Center for Medical Home Initiatives Web site ([www.medicalhomeinfo.org/screening/index.html](http://www.medicalhomeinfo.org/screening/index.html)).

### Future Efforts

Undoubtedly, the newborn screening efforts of the AAP will continue in the future. The year 2005 signified a great year of momentum for the AAP in the domain of newborn screening. In January, the BOD identified newborn screening as one of the year's primary initiatives and held a major-issue discussion at the face-to-face meeting. Throughout that discussion, the BOD con-

firmed the need for the AAP to assume a leadership role in the newborn screening initiative and outlined potential action steps. After that discussion on January 29, 2005, the BOD offered the following position statement.

The American Academy of Pediatrics supports the need for national leadership and consistency in the screening, diagnosis, and treatment of genetic diseases. The Academy commends the Secretary of Health and Human Services for efforts to achieve this goal and pledges to work collaboratively to see that genetic programs are based on the best scientific evidence. State programs should unify both public and private sectors to assure reliable and comprehensive screening and to form a seamless program to bring the best care available to children with genetic diseases—coordinated through the medical home, including appropriate pediatric subspecialists and the child's family.

On May 5, 2005, the BOD officially endorsed the ACMG report, which recommended states' implementation of a uniform screening panel. The endorsement highlighted the medical home's critical role in the follow-up process and pledged collaboration with other national partners, such as the March of Dimes, in promoting states' and practices' adoption of the report's recommendations. The AAP called for increased efforts in educating child health professionals to provide quality care for identified newborns, with the assurance that the AAP will use appropriate resources for the benefit of its 60 000 members.

The AAP will continue to foster national partnerships to engage in and promote the development of necessary programmatic and clinical standards for state-based programs and community-based practices. Collaborative discussions with the ACMG regarding opportunities to integrate genetic expertise into accessible and concise resources for child health professionals caring for children with positive newborn screen results, such as the ACMG action sheets, continue. Continued partnership with the CDC will allow for educational programming and resource expansion in the area of surveillance and screening in the medical home. Access to HRSA/MCHB-funded NNSGRC and the HRSA/MCHB-funded regional collaboratives will provide opportunities for increased technical assistance to child health professionals, families, and the general public, as well as the geographical sites to evaluate the pertinent clinical guidance of the AAP. Through these partnerships, the AAP intends to continue to assist child health professionals in their implementation of evolving national standards.

Electronic health records (EHRs) clearly have a role in the administration of quality health care. Their capacity to be a vehicle for providing medical homes to all children and their families demands their incorporation into all future screening activities of the AAP. With that in mind, HRSA/MCHB and the AAP convened a meeting in September 2004 to discuss strategies for reflecting the



pediatric population's needs in EHR standard setting, functionality, implementation, and future research. Participants at that meeting generated a list of recommendations, which was sent to the AAP BOD for consideration in 2004.

In January 2005, the AAP BOD approved a new program to operationalize the content of the AAP policy statements, clinical reports, and guidelines for future absorption into EHRs. The Partnership for Policy Implementation (PPI) cements the commitment of the AAP to contribute its intellectual capital comprehensively to standards-development efforts, which will enable EHRs to meet the needs of the pediatric community. The BOD acknowledges the role of AAP policy statements, clinical reports, and guidelines, ie, not only to serve as a premiere source of clinical guidance but also to communicate a minimal set of technical requirements for EHR developers. The establishment of these requirements will thus assist in ensuring that EHRs naturally automate the standard of care prescribed by the AAP.

PPI has 2 main objectives, ie, (1) to ensure that AAP policy statements, clinical reports, and guidelines are written so that EHR developers are able to incorporate them into decision-support systems and (2) to ensure that AAP policy statements, clinical reports, and guidelines are "implementable" in pediatric practice. To attain these objectives, PPI targeted 12 policy statements, clinical reports, or guidelines in its first year. Two consultants, namely, a pediatric informatician and a pediatric primary care quality improvement expert, were assigned to each targeted document. Through their consultation, the AAP's topical experts will develop clinical content that uses well-defined, unambiguous, standardized language and is clear in the presentation of the content's evidence base. Enabling the content to be explicitly written provides developers, standards-developing organizations, and other stakeholders with much-needed pediatric concepts for later absorption.

PPI will also develop an "implementation" section to be part of all targeted policy statements, clinical reports, and guidelines. This component will outline strategies for child health professionals to use to provide the standard of care delineated in the content. Embedding this information within the content enables the intellectual capital of the AAP to be a source of implementable clinical guidance.

MCHB is a partner in PPI, because the outcomes will assist in linking state newborn screening programs with medical homes. Defining these technical standards will lead to development of data and communication standards and ultimately construction of a common language for information management systems to be interoperable, allowing public health and practice-based systems to communicate with one another. These efforts ensure that the EHR framework supports the medical home approach to pediatric care, facilitating the provi-

sion of coordinated comprehensive care to all children and their families, which is a strategic priority of the AAP and HRSA/MCHB.

## REMAINING CHALLENGES

### Decision-Making Process Regarding Addition or Deletion of Conditions in Newborn Screening Panels

There is a paramount need to establish protocols for maintaining a uniform newborn screening panel. Currently, state and federal policies to determine which conditions are screened for are guided by inequitable variables, such as the evolution of treatments available for genetic/metabolic disorders or the presence of strong consumer advocacy groups for specific congenital disorders. For an effective, dynamic, decision-making matrix to be established, state policymakers, families, child health professionals, and newborn screening programs need to reach an ultimate consensus, despite the present context of disproportionate alignment of resources. The Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children can play an important role in this process.

### Communicating Results of Predictive Testing or Risk

Communicating screening results is a tender science. With the past screening paradigm, an infant is either identified as having a condition or not having a condition. However, with the emergence of new DNA-based technology, screening results now are predictive in nature, indicating a subject's higher risk for a congenital condition. This enhancement in the screening paradigm is one that calls for great attention to the communication practices of all health care professionals involved in the newborn screening process. As one mother indicated, "Consumers do not experience the test, diagnosis, the day-to-day struggles, on a population level; it is completely personal."<sup>14</sup> The implications for provider communication normative practices should be considered in future educational endeavors.

### Screening Newborns for Untreatable or Non-Life-Threatening Diseases

The idea that early health promotion activities in infancy or childhood can prevent later adult disease may cause a shift from screening newborns for diseases that are life-threatening or cause considerable morbidity to screening for diseases that are considered to have later onset, such as type 2 diabetes mellitus, heart disease, or cancer. Families report seeing benefits in newborn screening even if it identifies a condition for which no treatment is yet available. Parents also report their desire to know about genetic conditions in their family, so that they can make informed decisions about lifestyle (for the family and the child), choices of caregivers and specialists, financial planning, family planning, choices of jobs, edu-

cational choices, finding a support group, securing insurance, aiding in building registries, and participating in research. Both perspectives have additional implications for future screening programs and mandate that families, policymakers, and child health professionals reach consensus in defining the scope of this expanded system.

### Achieving Equity Among State Programs

Inevitably, tension may arise from federal efforts to achieve equity, either because of a perceived unfunded federal mandate or because of a state's inability to respond to a federal recommendation. We are committed to ensuring that federal efforts will be appropriately responsive and adequately financed to provide public health oversight of newborn screening programs while encouraging equity and innovation in the use of technology.

### CONCLUSIONS

The dynamic nature of state-based newborn screening programs will continue into the future, as the evolution of technology and communication methods continues. Federal and nongovernmental organizational efforts to address issues regarding newborn screening through the development of educational materials, standards, and resources and in partnerships with families are framed with the intent of ensuring a consistent coordinated system of care for newborns and children. It is this intent, with the desire to serve as a voice for all children and their families, that will continue to frame AAP and HRSA/MCHB pursuits in the future.

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