Serving the Family From Birth to the Medical Home

Newborn Screening: A Blueprint for the Future

Executive Summary: Newborn Screening Task Force Report

Approximately 4 million infants are born yearly in the United States (US), and are screened to detect conditions that threaten their life and long-term health. Newborn screening is a public health activity aimed at the early identification of infants who are affected by certain genetic/metabolic/infectious conditions. Early identification of these conditions is particularly crucial, as timely intervention can lead to a significant reduction of morbidity, mortality, and associated disabilities in affected infants.

Newborn screening has been universally accepted for the past 3 decades. It represented the first population-based genetic screening program, and signaled the integration of genetic testing into public health programs. Today, advances in technology are making possible new forms of newborn screening programs, such as newborn hearing screening. These technological advances will continue to have a significant impact on the sensitivity, specificity, and scope of newborn screening programs, including newborn heelstick screening.

Challenges are anticipated with technological advances. It is likely that public pressure to deploy new diagnostic capabilities, such as DNA-based technology, will increase despite limited knowledge of potential risks and benefits. In addition, the ability to detect individuals with conditions for which there is no effective or necessary treatment is likely. Further, as the Human Genome Project is completed, the impetus and opportunity for the transition of genetic technology into practice will increase. These and other challenges will affect not only newborn screening tests, but also the entire newborn screening system, which includes short-term follow-up, diagnosis, treatment/management, and evaluation. Inherent to each of these components is an education process. A national dialogue and process is needed to support state newborn screening systems as they try to keep pace with new technology.

To address these and other issues, a national Task Force on Newborn Screening (Task Force) was convened by the American Academy of Pediatrics (AAP) with funding from and at the request of the Maternal and Child Health Bureau (MCHB), Health Resources and Services Administration (HRSA), US Department of Health and Human Services (HHS). The AAP was asked to convene the Task Force in recognition that pediatricians and other primary care health professionals must take a lead in partnering with public health organizations to examine the many issues that have arisen around the state newborn screening programs.

To ensure that children who are screened are linked to a medical home, it was essential that pediatricians and other primary care health professionals be involved. The AAP defines the medical home as care that is accessible, family-centered, continuous, comprehensive, coordinated, compassionate, and culturally competent. A child who has a medical home has a pediatrician or other primary care health professional who is working in partnership with the child's family to ensure that all medical, nonmedical, psychosocial, and educational needs of the child and family are met in the local community.

Task Force members were appointed to represent many perspectives among those who operate programs, conduct research, and are affected by newborn screening systems. The co-sponsors of this effort were: other HHS agencies including the National Institutes of Health (NIH), the Centers for Disease Control and Prevention (CDC), and the Agency for Healthcare Research and Quality (AHRQ); the Genetic Alliance, which is a consortium of consumer groups; and national public health organizations including the Association of State and Territorial Health Officials, the Association of Maternal and Child Health Programs, and the Association of Public Health Laboratories. This report has been approved by the AAP Board of Directors. It does not necessarily reflect the viewpoints of sponsoring organizations or the organizations represented by members of the Task Force.

The purpose of the Task Force was to review issues and challenges for state newborn screening systems. The review process was structured to further expand representation. Task Force members were divided into 5 work groups, and additional individuals were invited to participate in each work group's examination of key issues. Over the course of 6 months, questions, concerns, and issues were collected from state public health agencies, state public health laboratory directors, maternal and child health programs, pediatricians, and other primary care health professionals who care for children, families and other consumers, bioethicists, scientists, and health services researchers. Each work group formulated conclusions and developed consensus recommendations. On May 10–11, 1999, the Task Force heard presentations from the 5 work groups, along with public comment on the reports and recommendations. A set of recommendations was developed incorporating key elements of the work group reports, issues raised by the public, and other related information. This document summarizes the Task Force recommendations.

The Task Force has outlined a national agenda for strengthening each "state" newborn screening system. ("State" newborn screening systems refer to state and territorial programs for heelstick newborn screening.) The Task Force believes that public health agencies (federal and state), in partnership with health professionals and consumers, should continue to:

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

KEY RECOMMENDATIONS

I. Effective Newborn Screening Systems Require an Adequate Public Health Infrastructure and Must Be Integrated With the Health Care Delivery System

- Federal agencies must take action to strengthen the public health infrastructure for newborn screening.
  - — The federal government—acting through the HRSA, CDC, Health Care Financing Agency (HCFA), AHRQ, NIH, and other agencies—should collaborate to provide ongoing leadership and support for development of newborn screening standards, guidelines, and policies.
  - — As the federal unit with most responsibility for newborn screening system development, the HRSA should engage in a national process involving government, professionals, and consumers to advance the recommendations of this Task

Force and assist in the development and implementation of nationally recognized newborn screening system standards and policies.
  - — Federal resources should be identified to sustain a Newborn Screening Quality Assurance Program to assist state public health laboratories. Such assistance must be both sustained and expanded as states adopt new screening technologies and modalities.
  - — The HRSA's MCHB should strengthen current mechanisms to improve coordination of infant health programs and initiatives within the state and/or between states, including continuation of funding in support of newborn screening program reviews.
- State public health agencies should direct their newborn screening program to be consistent with professional guidelines and recommendations. Each state public health agency should take responsibility for systems development. Specifically, states and their agencies have responsibility to:
  - — Design and coordinate the newborn screening system;
  - — Adhere to nationally recognized recommendations and standards for the validity and utility of tests. State newborn screening systems have a responsibility to review the appropriateness of existing tests, tests for additional conditions, and new screening technology and modalities; and
  - — Adopt standards for laboratories, health professionals, and health care financing plans based on nationally recognized standards and guidelines for follow-up, diagnosis, and treatment.
- State public health agencies, working under legislative authority, have the ongoing responsibility to ensure quality and evaluate program effort. States and their state public health agencies should:
  - — Maintain a newborn screening system that has appropriate evaluation, performance monitoring, and quality assurance activities from initial screening, through follow-up, diagnosis, treatment, and services through adolescence and adulthood;
  - — Conduct oversight of program operations, including those outside the public health agency, such as test analysis and tracking, private sector collection and transmission of screening data, laboratory quality, and the quality of the diagnostic procedures and treatment programs at pediatric subspecialty clinics; and
  - — Monitor and evaluate program performance through collection, assembly, analysis, and reporting of data, including outcome evaluations.
- States and state public health agencies should implement mechanisms to inform and involve health professionals and the public. Each state should:
  - — Develop a program advisory board that is multidisciplinary, involves pediatricians and other primary care health professionals who provide medical homes for children, pediatric subspecialists, and has meaningful representation of families and the general public; and
  - — Design and implement public, professional, and parent education efforts regarding newborn screening.
- States and state public health agencies should provide support for coordination and integration of program activities, including information and services. This will require public—private, federal—state, and intrastate partnerships. States should:
  - — Use public and private resources to fund demonstration programs that can serve as a testing ground for linking information and services in ways that improve the newborn screening system; and
  - — Structure interagency coordination to maximize resources and to improve the efficiency and effectiveness of newborn screening systems.

II. Public Health Agencies Must Involve Health Professionals, Families, and the General Public in the Development, Operation, and Oversight of Newborn Screening Systems

- The pediatrician or primary care health professional who, in partnership with parents, is the source of the child's medical home, should:
  - — Ensure that all newborns admitted to their practice have received adequate newborn screening, and that appropriate documentation of testing is present;
  - — Follow positive screening results to diagnosis (ie, confirmed or excluded), including repeated screening and diagnostic testing;
  - — Coordinate a seamless system of care with pediatric subspecialty clinics, tertiary care centers, and/or community-based providers, when a child is diagnosed with a disorder through newborn screening;
  - — Maintain a central record and database containing all pertinent medical information about the child. This record should be accessible to the family and others involved in the child's care, but confidentiality must be ensured; and
  - — Assist the family in understanding the diagnosis, symptoms, and potential implications of a diagnosed genetic/metabolic condition, as well as the availability of genetic counseling, family testing, and other family support services.
- Parents should receive information (on behalf of their children) about newborn screening.
  - — Prospective parents should receive information about newborn screening during the prenatal period. Pregnant women should be made aware of the process and benefits of newborn screening and their right of refusal before testing, preferably during a routine third trimester prenatal care visit.
  - — Parent knowledge should be reinforced after delivery by educational materials and discussion as needed by the infant's pediatrician or primary care health professional and/or knowledgeable hospital staff.
  - — Prenatal health care professionals as well as the infant's primary care health professional should be knowledgeable about their state's newborn screening program through educational efforts coordinated by the state's newborn screening program in conjunction with a newborn screening advisory body.
- Written documentation of consent is not required for the majority of newborn screening tests, for example, those tests of proven validity and utility.
  - — Parents should always be informed of testing and have the opportunity to refuse testing.
  - — If after discussions about newborn screening with health professionals, parents refuse to have their newborn tested, this refusal should be documented in writing and honored.
  - — If a newborn screening test is investigational or in the process of being developed, the benefits or potential risks have yet to be demonstrated, and identifiers are not removed from the specimen, informed consent should be obtained from parents and documented.
- Studies should be performed to broaden understanding of the ways in which communication can be performed more effectively for the benefit of consumers.
  - — Pilot studies and evaluation research should be conducted to assess the potential impact of revised parental permission and informed decision-making policies.
  - — Each state or region should, with input from families who have children with special needs and/or parent information centers, develop and provide family educational materials about newborn screening.
  - — Evaluation of materials should be ongoing, particularly because of the changing demographics of childbearing, cultural changes, and rapid developments in genetic science.
- Parents have a right to confidentiality and privacy protections for the medical and genetic information in any type of newborn screening results. Based on nationally recognized standards and guidelines, each state should have appropriate policies and mechanisms in place to ensure families' privacy and confidentiality. Laws to guarantee genetic privacy and protect against genetic discrimination should benefit patients identified by newborn screening.
- States and the federal government should include public participation in medical policy-making. The Secretary's Advisory Committee on Genetic Testing provides a mechanism for public participation in genetic policy development at the federal level. Each state should establish and fund a newborn screening advisory body with public participation to advise on newborn screening policy developments.
  - — Such an entity should include a broad range of public advisors representing parents, health professionals, third-party

payers, appropriate government agencies, and other concerned citizens.
  - — Such an entity should be empowered to advise state officials about screening for particular conditions based on accepted standards and be consulted about the development of related state regulations.
  - — Such an entity should be involved in the review of new tests under consideration by the state and in the development of pilot programs for new tests.
  - — Such an entity should be involved in the ongoing evaluation of all aspects of the state's process for newborn screening. Oversight activities should include a review of: testing, follow-up, and treatment efforts; the impact on families of receiving a false-positive screening result; and the state's process for handling consumer input including grievances.

III. Public Health Agencies Must Ensure Adequate Infrastructure and Policies for Surveillance and Research Related to Newborn Screening

- State Maternal and Child Health (MCH) programs should conduct a review of the newborn screening system and its relationship to the HRSA MCH Block Grant Performance Measures and evaluate the quality of data of the newborn screening-related performance measures.
- The federal HCFA should develop Health Plan Employer Data and Information Set (HEDIS) measures to evaluate the health plans' performance within the newborn screening system.
- A federally-funded newborn screening research agenda should be outlined that aims to: develop better tests (more sensitive, more specific, and less costly); assess the validity and utility of new technologies (eg, tandem mass spectrometry, DNA-based testing, and other evolving technologies); and define appropriate uses of residual biologic samples for population-based research and surveillance.
- The HRSA's 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 data sets, analyses of cost-efficiency and effectiveness, and integration with other public health data systems. Support for technological innovation (ie, new test technologies) should include these measures.
- Pediatricians, pediatric subspecialists, and other health professionals who care for children should contribute to newborn screening data collection to advance knowledge about health outcomes and intervention effectiveness. Professional associations, the HRSA-funded National Newborn Screening and Genetics Resource Center, and state newborn screening programs should develop strategies to assist health professionals in their efforts to participate in and learn from newborn screening information systems.
- Pilot studies should be undertaken to demonstrate the safety, effectiveness, validity, and clinical utility of tests for additional conditions and new testing modalities. Informed consent of parents is called for in all such pilot studies. These studies might be undertaken by individual states, regional or nationwide groups of states, or through federal grants provided to research institutions across the country.
- Federal and state public health agencies, in partnership with health professionals, families, and representatives of ethnic, minority, and other diverse communities should:
  - — Develop model legislation and/or regulation that articulates policies and procedures regarding utilization of unlinked and identifiable residual samples for research and public health surveillance. This process should include review and consideration of the recent recommendations to the President set forth by the National Bioethics Advisory Commission (NBAC) for research involving human biological materials;
  - — Develop model consent forms and informational materials for parental permission for retention and use of newborn screening samples;
  - — Develop educational materials for parents that includes information regarding the storage and uses of residual samples;
  - — Organize collaborative efforts to develop minimum standards for storage and database technology to facilitate appropriate storage of residual newborn screening blood samples at the state level; and
  - — Consider creating a national or multi-state population-based specimen resource for research in which consent is obtained from the individuals from whom the tissue is obtained. Such a resource could be an alternative to retaining newborn screening samples for potential use in research.
- Using national recommendations, each State program should develop and implement policies and procedures for retention of residual newborn screening blood samples that articulate the rationale and objectives for storage, the intended duration of storage, whether storage is with or without identifiers, and guidelines for use of identifiable and unlinked samples. An advisory group for newborn screening programs with broad health professional and family/community representation is a valuable resource in developing policies and procedures and in reviewing applications for use of retained samples. The advisory body also could determine priorities for use.

IV. Public Health Agencies Should Ensure Adequate Financing Mechanisms to Support a Newborn Screening Program

- States should ensure adequate financing of all parts of the newborn screening system: screening, short-term follow-up, diagnostic testing, comprehensive medical care/treatment, and evaluation of the system. If newborn screening fees are not adequate, funding of all components of the system could be accomplished with other public health dollars or by third-party payers. Other uses of newborn screening fees should not be considered until all of the components of the newborn screening system are fully funded.
- States should take responsibility for blending resources available through Title XIX (Medicaid), Title V (MCH Block Grant), Title XXI (State Children's Health Insurance Program) [SCHIP], and private insurance to guarantee necessary coverage and financing for all children and adolescents with a condition diagnosed through the newborn screening system.
- State contracts for publicly-subsidized third-party insurance plans that cover children (eg, Medicaid and SCHIP) should explicitly require coverage for newborn screening and those services and treatment related to disorders identified by newborn screening. State contracts also should require that third-party payers ensure access to health care professionals with appropriate pediatric expertise within the network or through out-of-network referrals.
- States, in cooperation with health professionals and payers, should put mechanisms in place to identify the third-party payers for newborns immediately following birth. For example, all states should operationalize the automatic newborn eligibility requirements under Medicaid and the Health Insurance Portability and Accountability Act (HIPAA) newborn coverage provisions that require infant coverage and prohibit preexisting condition exclusions for newborns.
- Purchasers—public and private—should ensure that the benefits package they pay for includes the care and services defined by the AAP Scope of Health Care Benefits Statement and the Council of Regional Networks for Genetic Services Guidelines.
- In the Supplemental Security Income (SSI) program, the federal government should review the technical appropriateness of guidelines, and evaluate the consistency of their application, for children with conditions identified through newborn screening.

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