

Newborn Screening Position Statement

ASTHO affirms that early identification, diagnosis, and treatment of infants with disorders identified during newborn screening (NBS) are vital to preventing significant morbidity and mortality and the retaining of residual blood spot cards from these tests is essential for future disease prevention. Each year, over four million newborns in the United States are screened for a variety of genetic and other congenital disorders. Newborn Screening tests help to prevent or limit the medical consequences associated with metabolic, endocrine, hemoglobin, infectious, hearing, and other generally asymptomatic conditions.

In 2008, the Newborn Screening Saves Lives Act (S. 1858) established grant programs to provide education and outreach on NBS and coordinated follow-up care. It asked the CDC, HRSA and state health agencies to develop a national contingency plan for NBS programs in the event of a public health emergency. It also authorized funding to enhance or expand the ability of state and local public health agency NBS programs to evaluate their effectiveness and provide quality assurance for NBS laboratories. While the American College of Medical Genetics (ACMG)/Health Resources and Services Administration (HRSA) issued guidelines for NBS programs in 2005, states have independently set policies for their programs and vary in their capacity to execute them.

State public health agencies have been administering NBS programs for over 40 years through the use of a dried blood spot card collected from the heel of each newborn. In the short-term, these blood spots are essential for screening infants. In the long-term, they are necessary for evaluating current NBS tests and developing new ones (74% of states), clinical and forensic testing (52%), and epidemiologic studies (28%). Currently, 10 states keep blood spot cards indefinitely. Many states retain them short-term.

ASTHO's Principles for a Comprehensive Newborn Screening System

- NBS is a core state public health service, and should be mandatory. As a comprehensive system, any mandated NBS panel must be accompanied by adequate resources to ensure that screening, follow-up, diagnosis, treatment, and management services occur, regardless of an individual family's ability to pay.
- NBS is critical to the early identification, diagnosis and treatment of infants thus vital to preventing significant morbidity and mortality and should be a mandatory benefit provided for every child.
- NBS and follow-up services must be provided in community-based settings close to the individuals and families receiving services and tailored to the particular needs of each community.
- Information on newborn screening services, screened conditions, options, and technologies must be presented in an unbiased manner and be culturally and linguistically appropriate.
- Systems must be accessible and must incorporate evidence-based testing and treatment, as well as funding that balances the interests of the affected newborn with those of society.
- The NBS, Hearing Screening Program, Vital Records, and the Birth Defects Surveillance System must work together, and to the extent possible link NBS data with these other birth data.
- To the extent feasible, NBS, including hearing, results must be included in electronic health records to enhance surveillance and short-term follow-up.
- Decisions regarding NBS panels must consider: burden of illness; birth prevalence of the disorder in the population to be screened; whether the disorder can be detected with a screening test that is ethical, safe, accurate, and cost effective; the impact of false positive tests on families; the availability of effective treatment for the disorder and whether early treatment (i.e., before the onset of symptoms) is more effective in improving health outcomes than later treatment; and the ability to finance all components of the NBS program, not only laboratory testing.

- Evolving technology may make it possible to test for conditions for which the etiology of the condition is unknown and for which there is no known effective treatment. Laboratories may choose to offer testing for these conditions. In such circumstances, parents and health care providers have the responsibility to determine the best course of care.
- Long-term follow-up begins once treatment for a condition starts. Follow-up ends when the youth transitions into an adult treatment and service delivery system at 21 years of age. However, adult service delivery systems must work with pediatricians to address the ongoing needs of transitioning youth so that their health is not jeopardized by lack of access.
- NBS programs must represent a comprehensive and coordinated system encompassing education, screening, follow-up, diagnosis, evaluation, and management.
- Coordination must occur with private insurers, SCHIP, and Medicaid to arrange insurance coverage for newborn screening and those with screened conditions.
- Health information technology and exchange, to support immediate and long-term follow-up and to reduce errors in providing healthcare must be used whenever feasible. There must be transparency in the transfer of information back to labs to prevent loss to follow-up and late results.
- Children with disorders must have a “medical home” in which clinicians provide integrated healthcare services that both address the patients’ healthcare needs and promote partnerships between providers and patients.

State and Territorial Health Agency Role

- Ensure that there are sufficient personnel for sample analysis and short-term follow-up. Strategies for assuring adequate expertise in urban and rural areas and multi-lingual communities are particularly important. An adequate health care and public health workforce is critical to the expansion and long-term management of any NBS program.
- Provide leadership in promoting federal, state, territorial, tribal, and local solutions to ensure affected children receive the necessary follow-up in the absence of a universal and comprehensive, national long-term follow-up system.
- Adopt and incorporate the ACMG/HRSA recommended uniform panel of all core conditions.
- Develop contingency plans at every level in the event of a public health emergency. States must support effective and timely reporting of screening results to physicians and families and continuing diagnostic confirmation of positive screening results. The availability of treatment and management resources must be ensured, and families should be educated about NBS in the event of a state or national emergency.
- Maximize service coordination at state and local levels as well as program integration with the State Children’s Health Insurance Plan, early intervention programs, Title V programs, and similar services. Failure to consider the system in its entirety can result in a fragmented system unable to improve the public’s health.
- Integrate NBS data systems with vital registration, immunization registries, birth defects registries, NBS short-term follow-up, and genetics databases to promote efficiency and continuity of care.
- Work with medical associations, hospitals, family groups, and others to increase public awareness and improve health literacy about NBS programs and to ensure that professional training and family education reflects current and accurate health information.

- Continue to facilitate the development of healthcare systems in which primary care clinics serve as a “medical home,” provide or link affected children to needed personal health services, and monitor the overall health status of the patients under their care.
- Participate in regional and national efforts to collect and monitor information on long-term outcomes for infants with positive screening results and confirmed disease, as infrastructure and program resources are available.
- Develop policies that incorporate best practices from state health agencies across the nation, that articulate the rationale, objectives, and procedures for retaining NBS residual bloodspots. These policies should provide guidelines on duration of storage, use of identifiable and unlinked samples, security and confidentiality of the samples, and allow for individuals to opt-out of blood-spot storage.
- Periodically review and update state plans to assess effectiveness and reflect changes in science and technology.

Federal Government Role

- Consider long-term follow-up efforts a national activity. The federal government must address access to affordable, coordinated health services when affected children move between states or transition into adult care.
- Provide resources to increase state capacity to administer comprehensive NBS systems, share effective practices, and provide a forum to conduct ongoing evaluation of any recommended panel of tests or emerging technologies.
- Support state laboratories and public health agencies that provide NBS services during emergency situations with guidelines, technical assistance, and other resources.
- Establish standards for data collection:
 - Linking data systems such as Vital Records and Birth Defects Surveillance with NBS;
 - Promoting data sharing and collaboration at the federal, regional, state, territorial, tribal and local levels to develop guidelines on long-term follow-up and treatment;
 - Tracking individuals as they move geographically and through systems; and
 - Providing assistance to states to carry out standards.
- Develop a mechanism, consistent with state and federal confidentiality laws, to provide state newborn screening programs or researchers access to information regarding location and availability of newborn blood spot repositories.
- Provide technical assistance to states regarding interpreting privacy regulations, confidentiality concerns, prevention of discrimination, and quality assurance associated with NBS.
- Maintain an adequate safety net for children with heritable disorders and genetic diseases, who are often not considered “disabled” for the purposes of Medicaid.



POSITION STATEMENT

Approval History

Policy Committee Review and Approval on July 31, 2009

Executive Committee Review and Approval on August 28, 2009

Ratified by the ASTHO Membership on September 29, 2009

Policy Expires on September 29, 2012

ASTHO policies are broad statements of enduring principles related to particular policy areas that are used to guide ASTHO's actions and external communications.

Related ASTHO Documents

- ASTHO General Policy Statement
- Access Policy Statement