Newborn Screening Policy Statement

Position:

ASTHO recognizes and affirms that newborn screening (NBS) is a core public health service that is vital to preventing significant infant morbidity and mortality through early identification of heritable, congenital, and inborn-metabolic conditions.

Summary of Recommendations:

- Establish clear guidelines on how specimens will be transported, stored, and used in research. Assure programs meet national guidelines.
- Ensure an adequate healthcare and public health workforce to support NBS activities.
- Facilitate access to accurate and easy-to-understand educational materials in multiple languages for families.
- Collaborate with partners to ensure diagnostic and treatment services are accessible through a medical home model.
- Support and track short- and long-term follow-up of testing and diagnosis of disease based on screening results.
- Coordinate with public and private insurers to arrange insurance coverage for diagnosis and long-term treatment of inborn metabolic and congenital conditions.
- Use health information technology and exchanges when feasible to link data.

Background:

For more than 50 years, state and territorial public health agencies have instituted NBS programs to detect infants at risk of heritable, congenital, and inborn-metabolic conditions using bloodspot samples, pulse oximetry, and hearing screening of nearly all four million babies born each year in the United States.[1] NBS is non-diagnostic, but rather a screening, and informs providers of the need for further testing. Approximately 12,900 babies requiring further testing are identified each year through NBS. That opportunity for early intervention yields lifetime benefits for children, their families, and health systems by preventing significant morbidity and mortality.[2, 3]

The Newborn Screening Saves Lives Reauthorization Act of 2014 (NBSSLRA) amended the Public Health Service Act to reauthorize federal programs through 2019 that support states and territories in improving and expanding NBS programs.[4] NBSSLRA directs activities across various agencies within HHS to improve the quality and capacity of state NBS programs. The Act reauthorized federal programs that support provider and parent education, ensure laboratory quality, and increase surveillance for NBS programs guided by the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC).[5] The NBSSLRA legislation is currently pending in Congress to reauthorize newborn screening programs.[6]

All jurisdictions (including Washington, D.C., Puerto Rico, Guam, and the Virgin Islands) screen for at least 30 of the 35 core and 26 secondary conditions.[7, 8, 9] Unfortunately, as new conditions are added to state NBS panels, laboratories may experience significant challenges incorporating new, costly technologies and the associated complexity of routine data analysis necessary to identify at-risk newborns. States may also grapple with implementing systems for follow-up and care management for increasingly complex conditions added to state NBS panels.

Recommendations and Evidence-Base:

ASTHO recommends the following policy considerations for NBS programs:

- When deciding on NBS panels, states and jurisdictions are encouraged to follow the same principles as the ACHDNC.[10] The ACHDNC issues the Recommended Uniform Screening Panel, a national guideline for NBS consisting of conditions for which the Secretary of HHS provides guidance.
- Establish clear guidelines regarding length of time specimens will be stored; the rationale, objectives, potential risks or benefits, and procedures associated with NBS dried bloodspot retention and research; use of identifiable and unlinked samples; security, privacy, and confidentiality of the samples; the conditions of storage; and allowances for parents or the child upon turning 18 years old to opt out of bloodspot storage or use of their bloodspots for future research.[11]
- Support of a robust healthcare and public health workforce to manage the NBS system comprised of sample collection, screening, education, system quality assurance, medical management, and periodic outcome evaluation. This includes hospital and birth centers, sample transport networks, NBS and diagnostic laboratories, short term-follow up services, and primary and specialty healthcare providers.
- Collaborate with medical associations, healthcare facilities, policy makers, and family groups to improve access to clear, accurate, and easy to understand information in multiple languages on NBS and dried blood spot storage, and further diagnostic testing.[12,13]
- Provide a pathway to ensure short- and long-term follow-up once diagnosis occurs, considering the life course of the child, the natural history of the condition, and its long-term physical, social, and economic effects.
- Participate in regional and national efforts to collect and monitor long-term outcomes for infants with positive screened results and confirmed disease, including access to treatment and support.
- Coordinate with public and private insurers to cover NBS and diagnostic testing and ensure that a safety net exists for children without insurance. Safety net services include coverage for prescribed food, medication, or services required to prevent disability or death in children with disorders detected through NBS.
- Build and manage collaborative treatment and support services that are accessible to children with diagnosed disorders through a medical home model. Support funding for integrated healthcare services, including expanding access to telemedicine clinical and support services.
- Link children and families with diagnosed disorders to Title V Children and Youth with Special Health Care Needs Programs, hospitals, education, social services systems, mental and behavioral health supports, and Special Supplemental Nutrition Program for Women, Infants, and Children (WIC) programs.
- Link NBS data to death records prior to follow-up with parents to prevent contact if an infant is recently deceased.[14] To the extent possible, ensure interoperability, or secure two-way data access and exchange, between NBS, point-of-care screening, Vital Records, Birth Defects Surveillance, and Pregnancy Risk Assessment Monitoring systems.[15]
- Use health information technology and health information exchanges whenever feasible to support follow-up services and reduce diagnostic and treatment errors and incompleteness.
- As resources allow, participate in national and state efforts to collect, monitor, and develop consensus around standard aggregate quality indicators or metrics representing all infants screened. Standard indicators help public health agencies monitor short-term outcomes for infants with positive screening results and to collect and monitor NBS data to understand problems, implement quality improvement activities, and monitor progress (e.g., the Newborn Screening and Technical Assistance Evaluation Program (NewSTEPs)).[16]
- Assure that, to the best of their ability, state and territorial NBS programs meet national guidelines by updating plans, processes, and procedures, including appropriately reacting to changes in science

and technology, incorporating recommended screening condition, and ensuring the timeliness of screening and associated diagnoses.

- Develop contingency plans at every level in the event of a state, national, or laboratory emergency to support effective and timely collection and testing of samples, reporting of screening results to physicians and families, and continued diagnostic confirmation of positive screening results. This includes making plans to support the availability of treatment and management resources, including funding and integrating long-term access to telemedicine services.

Approval and Revision Dates

Community Health and Prevention Policy Committee Approval: August 6, 2021
Board of Directors Approval: October 20, 2021
Policy Expires: October 31, 2024

ASTHO membership supported the development of this policy, which was subsequently approved by the ASTHO Board of Directors. Be advised that the statements are approved as a general framework on the issue at a point in time. Any given state or territorial health official must interpret the issue within the current context of his/her jurisdiction and therefore may not adhere to all aspects of this Policy Statement.

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[2] U.S. Department of Health and Human Services. "2015 Report to Congress: Newborn Screening Activities." Available at https://mchb.hrsa.gov/sites/default/files/mchb/MaternalChildHealthTopics/Perinatal/NBS%20Report.pdf. Accessed 4-1-2019.

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[5] Health Resources and Services Administration (HRSA). "Considerations and Recommendations for National Guidance Regarding the Retention and Use of Residual Dried Blood Spot Specimens After Newborn Screening." 2010. Available at https://www.hrsa.gov/sites/default/files/hrsa/advisory-committees/heritable-disorders/reports-recommendations/reports/briefing-residual-dried-spot-specimens.pdf. Accessed 2-4-2019.

[6] H.R.482 - 117th Congress (2021-2022):" Newborn Screening Saves Lives Reauthorization Act of 2021". Available at https://www.congress.gov/bill/117th-congress/house-bill/482. Accessed 05-24-2021.

[7] NewSteps. "Newborn Screening Status for all Disorders. Available at https://www.newsteps.org/resources/data-visualizations/newborn-screening-status-all-disorders. Accessed 5-5-2021.

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[10] Kemper AR, Green NS, Calonge N, Lam WK, Comeau AM, Goldenberg AJ, Ojodu J, Prosser LA, Tanksley S, Bocchini JA. "Decision-making process for conditions nominated to the Recommended Uniform Screening Panel: statement of the US Department of Health and Human Services Secretary's Advisory Committee on Heritable Disorders in Newborns and Children." *Genetics in Medicine*.

[11] Health Resources and Services Administration (HRSA). "Considerations and Recommendations for National Guidance Regarding the Retention and Use of Residual Dried Blood Spot Specimens After Newborn Screening." 2010. Available at https://www.hrsa.gov/sites/default/files/hrsa/advisory-committees/heritable-disorders/reports-recommendations/reports/briefing-residual-dried-spot-specimens.pdf. Accessed 2-4-2019.

[12] Botkin JR,et al. Prenatal Education of Parents about Newborn Screening and Residual Dred Blood Spots A Randomized Clinical Trial. *JAMA Pediatrics*. 2016;170(6):543-549. 2013. Available at http://www.hrsa.gov/advisorycommittees/mchbadvisory/heritabledisorders/reportsrecommendations/reports/decisionmakingprocessforconditions.pdf. Accessed 2-1-2019.

[13] American Academy of Pediatrics. Newborn Screening Authoring Committee. Newborn Screening Expands: Recommendations for Pediatricians and Medical Homes – Implications for the System. Pediatrics 2008; 121(1): 192-205

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[15] Office of the National Coordinator for Health IT. "Interoperability." 2019. Available at https://www.healthit.gov/topic/interoperability. Accessed 9-24-2020.

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