Uses of Newborn Screening Residual Dried Blood Spot Specimens and Test Results

APHL POSITION STATEMENT

Statement of Position

Public health newborn screening (NBS) programs should have appropriate policies for retention and use to allow for storage and use of residual dried blood spot (DBS) specimens and test results for program activities without requiring parental consent.[1]

Recommended by: **APHL Newborn Screening Committee**
Approved by Board of Directors: **October 2025**
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Sunset Date: **November 2030**

Contact: Amanda Cosser
Manager, Public Policy & Regulatory Affairs
240.485.2324 or amanda.cosser@aphl.org

Background

NBS is a program that screens newborns at birth for a select number of serious diseases that, if left undetected and untreated, could result in morbidity and/or mortality. NBS is mandatory in all US states and territories (henceforth, "states"), and each state has its own NBS panel, which may vary in the number of conditions that are screened.

NBS is performed on DBS specimens collected via heel stick. These specimens are tested for congenital diseases and newborns with actionable results receive follow-up services. NBS follow-up is defined as "actions taken to ensure that a newborn whose specimen is unacceptable, or whose screening result warrants additional action receives appropriate repeat screening, diagnostic testing, clinical evaluation and/or intervention."[2]

The Association of Public Health Laboratories (APHL) endorses the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) report: *Considerations and Recommendations for National Guidance Regarding the Retention and Use of Residual Dried Blood Spot Specimens after Newborn Screening, 2011,*[1] which states that NBS programs should have a retention and use policy in place that has been reviewed by a state attorney general or other legal authority. This policy should specify who is responsible for storage, access, use and secure destruction of residual DBS specimens after testing is completed.[1] Additionally, states should consider incorporating language into their state policy to control access to residual DBS specimens by law enforcement. Sample language developed by APHL's NBS Ethical, Legal, Social and Policy Issues Subcommittee was presented at the 2024 NBS Symposium.

Once screening is complete, state programs remove identifying information from DBS cards and retain residual DBS specimens for various lengths of time, from several weeks to over 18 years.[1] These specimens and test results are used to support essential program functions such as program evaluation, quality assurance, result verification, test refinement and quality improvement initiatives.[1, 3-5] Use and analysis of residual DBS specimens and test results is essential for several NBS program activities, including:

- Laboratory quality control, quality assurance (e.g., validating screening results) and programmatic improvement
- Calibration of testing equipment
- Evaluation of equipment, reagents, and screening methods (and their development)
- Validation of equipment and screening methods
- Development, testing and maintenance of a continuity of operations plan to ensure testing can continue in the event of an emergency
- Assuring the competency of testing personnel.

Because NBS programs are state-mandated and involve regulated laboratories, program activities related to ensuring accuracy, quality and improvement of NBS tests are considered public health practice, not research.[6]

NBS activities may include studies for the development of new laboratory tests, which are essential for expanding public health's ability to protect newborns from congenital conditions where early detection is key. These studies may or may not be research, and opt-in or opt-out consent may be indicated, depending on the nature and details of the study, any applicable Institutional Review Board (IRB) determinations, the funding source and applicable state laws.

APHL Implementation Steps

1. APHL will share this position statement with key state and federal policy makers.
2. APHL will collect and maintain information on residual DBS specimen and test result retention times and uses. This information will be used to educate NBS program policymakers.
3. APHL will collaborate with the Centers for Disease Control and Prevention (CDC) and NBS partners to collect, evaluate and share best practices, model policies and parent/provider educational materials. This information will be used to develop guidance for NBS programs on secure storage and use of residual DBS specimens and test results.
4. APHL will encourage NBS programs to develop policies on retaining, storing and using residual DBS specimens and test results that are compliant with the recommended Clinical and Laboratory Standards Institute (CLSI) storage conditions and federal and state research privacy rules and regulations.[3,7]

Resources

1. Therrell BL, Hannon WH, Bailey DB, Goldman EB, Monaco J, Norgaard-Pedersen B, Terry SF, Harris A, Vasquez LM, Johnson A, Lloyd-Puryear MA, Howell RR. Committee report: Considerations and recommendations for national guidance regarding the retention and use of residual dried blood spot specimens after newborn screening. Genet Med. 2011 Jul; 13 Accessed December 10, 2024 from: https://www.hrsa.gov/sites/default/files/hrsa/advisory-committees/heritable-disorders/reports-recommendations/briefing-residual-dried-spot-specimens.pdf
2. CLSI. Newborn Screening Follow-up and Education; Approved Standard – Third Edition. CLSI document NBS02-Ed3. Wayne, PA: Clinical and Laboratory Standards Institute; 2023.
3. CLSI. Dried Blood Spot Specimen Collection for Newborn Screening Programs; Approved Standard—Seventh Edition. CLSI document NBS01-Ed7. Wayne, PA: Clinical and Laboratory Standards Institute; 2021.
4. American College of Medical Genetics. Position statement on the importance of residual newborn screening dried blood spots. Accessed December 10, 2024 from: https://www.acmg.net/PDFLibrary/NBS-Blood-Spot-Retention.pdf
5. Therrell BL, Hannon WH, Pass KA, et al. Guidelines for the retention, storage, and use of residual dried blood spot specimen after newborn screening analysis: statement of the Council of Regional Networks for Genetic Services, Biochem Molec Med 57: 116-124, 1996.
6. Federal Policy for the Protection of Human Subjects ('Common Rule'), 45 C.F.R. § 46.102 (2018).
7. CLSI. Managing Laboratory Records. 1st ed. CLSI guideline QMS26. Clinical and Laboratory Standards Institute, 2021.