Briefing Paper: Executive Summary

Considerations and Recommendations for a National Policy Regarding the Retention and Use of Dried Blood Spot Specimens after Newborn Screening

Newborn screening is a highly successful public health program that identifies rare genetic, metabolic, hormonal and functional disorders and assures early management and follow-up for those affected. Newborn screening is regulated and implemented by states, and each state has laws that either require or allow newborn screening. Newborn screening policies are usually implemented with input from multi-disciplinary advisory committees that include consumers. While state responsibility allows for local control and accountability, it also gives rise to wide variation in practices across the country.

All US newborn screening programs obtain dried blood specimens for laboratory testing. Portions of these specimens (residual specimens) are generally retained for some period of time after testing is complete. The primary justification for retained residual specimens is to benefit the child and family by documenting that a specimen was collected, received, and properly analyzed. Residual specimens may also be used for result verification, quality assurance activities (including new test validation). A collection of stored specimens is often referred to as a "biobank."

Newborn screening specimens are unique. They are usually the first blood specimen in a baby's life and they are collected on essentially all newborns. They provide critical information about risk for certain inherited conditions. They also have the potential to generate population-based knowledge that can improve the health of children, support families, and provide information critical to understanding the antecedents of adult diseases.

Residual specimen storage must assure that the confidentiality and privacy of families is respected and that the specimens are protected. Policies are needed in each state to promote public trust, emphasize transparency of administrative practices, and create supporting information that encourages informed public participation.

This is a guidance document and statement of the U.S. Secretary of Health and Human Services' Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC); roster of members available at http://www.hrsa.gov/heritabledisorderscommittee/governance/roster.htm). It is designed to review the issues facing newborn screening programs and to develop a national guidance policy for programs that retain and use dried blood specimens after newborn screening is completed.

CONCLUSION/RECOMMENDATIONS

Since the initial guidance for retention, storage of use of residual dried blood spots in 1996 [1], there have been noticeable improvements in policy development. In state newborn screening programs, there are currently two distinct philosophies regarding the storage and use of residual dried blood spots: 1) short-term storage (<3 years), presumably for program quality assurance and test improvement; and 2) long-term storage (> 18 years), presumably for public health research.

There is heightened awareness in the research and consumer communities concerning both the potential value of specimens and the potential privacy issues. Privacy issues are compounded by the lack of standardized consent policies across state programs, the lack of a universal legal definition of specimen ownership once the screening process is complete, and the lack of public awareness of newborn screening.

Because newborn screening is the only medical screening program that reaches the entire population of newborns, it is unique and the processes surrounding it must be carefully and thoughtfully approached. Residual blood specimens provide an excellent opportunity for storage and use in a biobank after screening is complete and the results have been validated. However, at the present time, this is a secondary purpose that may not have been adequately addressed in state law or policy. Therefore, residual specimen use in this way must be carefully considered anticipating both the potential benefits and risks. To assist in this process, the ACHDNC makes the following recommendations:

1) ***All state newborn screening programs should have a legally reviewed and accepted policy addressing the disposition of dried blood specimens remaining after newborn screening testing is complete and the screening results have been validated.*** Multidisciplinary input, including consumers, should be solicited and thoughtfully considered in developing such a policy. This specimen disposition policy should include the length of time for which specimens will be stored and storage conditions. Compliance with storage processes included in NCCLS/CLSI Standard LA4-A5 [2] or its current editions is recommended. Any data linkages should be carefully addressed and privacy and confidentiality assured. .

2) ***All state newborn screening programs should have a legally reviewed and accepted policy that specifies who may access and use dried blood specimens once they arrive at the state-designated newborn screening laboratory, including further access after newborn screening tests are completed.*** Multidisciplinary input, including consumers, should be solicited and thoughtfully considered in developing such a policy. This specimen access policy should include any uses prior to and after the newborn screening laboratory testing and validation process. If uses of dried blood spot specimens outside of newborn screening are allowed, then handling and disposition of the specimen should be addressed and privacy and confidentiality of any associated patient information assured.

3) ***As part of the educational process of the newborn screening system, all state newborn screening programs should maintain and distribute educationally and culturally appropriate information that includes basic information about the use or potential use of the dried blood specimens.*** Where long-term storage policies or other options exist relative to storage of residual dried blood spots, such information should be included in prenatal education materials.

4) ***All state newborn screening programs should work proactively to ensure that all women receiving prenatal care are educated about newborn screening.*** This activity should include appropriate steps to inform and train prenatal care providers regarding their educational responsibilities within the newborn screening system. Processes should be in place to evaluate the extent, timing and understanding of prenatal education with an eye towards educational program improvement.

5) ***If residual blood specimens are to be available for any process outside of the legally required newborn screening process for which they were obtained, an indication of the parents' awareness and willingness to participate should exist in compliance with federal research requirements (45CFR46).*** A consent (opt in) or a dissent (opt out) process may meet this requirement depending on purposes for which specimens will be used. The use of residual specimens for program evaluation (e.g. repeat testing as a quality check) or process improvement (e.g. non-commercial, internal program new test development) are valid components of the newborn screening system and, therefore, should not require additional consent.

6) Newborn screening programs should assess the utility of any additional consent/dissent process implemented in order to better address issues of storage and use of residual dried blood specimens. The federal government is encouraged to consider this as a priority and to provide funding for utility assessment projects over the next 5 years.

7) The federal government is encouraged to provide administrative support and funding to develop:

- Model consent/dissent processes on the use of residual specimens in newborn screening;
- Model educational programs for the general public on the importance of newborn screening and the potential uses of residual specimens to generate population based knowledge about health and disease;
- National data on the utility of any additional consent/dissent processes implemented relative to potential research uses of residual newborn screening specimens;
- Educational materials with facts about potential uses of residual newborn screening specimens for both consumers and prenatal healthcare providers.

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1. Guidelines for the Retention, Storage, and Use of Residual Dried Blood Spot Samples after Newborn Screening Analysis: Statement of the Council of Regional Networks for Genetic Services. Therrell BL, Hannon WH, Pass KA, Lorey F, Brokoff C, et al., Biochem Molec Med 1996;57:116-24.
2. Clinical and Laboratory Standards Institute (CLSI). Blood collection on filter paper for newborn screening programs; approved standard—fifth edition. CLSI document LA4-A5. Wayne, PA: Clinical and Laboratory Standards Institute; 2007.