

Newborn Dried Blood Spot Screening: Residual Specimen Storage Issues

AUTHORS: Bradford L. Therrell, Jr, MS, PhD, and W. Harry Hannon, PhD

National Newborn Screening and Genetics Resource Center, Austin, Texas

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Address correspondence to Bradford L. Therrell, Jr, MS, PhD, National Newborn Screening and Genetics Resource Center, 1912 W. Anderson Lane #210, Austin, TX 78757. E-mail: therrell@uthscsa.edu

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COMPANION PAPERS: Companions to this article can be found on pages 231 and 239 and online at www.pediatrics.org/doi/10.1542/peds.2011-0970 and www.pediatrics.org/doi/10.1542/peds.2011-2572.

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Traditional newborn dried blood spot screening (NDBS) has expanded in recent years to include upwards of 50 different conditions in many states. Once the screening tests are complete, the residual blood specimens (RBSs) that remain are most often available for quality assurance and public health research. The articles by Botkin et al¹ and Bombard et al² contained in this issue of *Pediatrics* investigate the attitudes of parents in both the United States ($n = 3855$) and Canada ($n = 66$) toward program policies that allow for the possible storage and secondary use of RBSs.

Figure 1 illustrates the variability in the United States regarding the length of time RBSs are currently stored. Currently, 14 programs save RBSs for ≥ 21 years, and these programs serve $\sim 45\%$ of all US births. Although the need for RBSs for laboratory quality assurance, which may include long-term needs, is generally understood (and accepted), the way in which programs disclose (or fail to disclose) information about these uses and the manner by which parents may choose to opt in or out of RBS storage and/or potential research uses are the subject of much discussion and controversy.

The Secretary of Health and Human Services' Advisory Committee on Heritable Disorders in Newborns and Children has recently recommended extensive educational efforts for parents and health care professional alike regarding the possibility of secondary uses of residual NDBS specimens.³ Although these recommendations focus primarily on improved education and transparent policies, they also suggest the possibility of a national RBS repository to which parents could direct their newborn's RBS. A virtual blood spot repository available to researchers and a data repository in which clinicians could deposit long-term follow-up data are already in development.⁴

It is important that primary care physicians educate themselves as to their role in the NDBS system, whether in the United States or Canada or elsewhere. The value of NDBS for improving health outcomes in newborns and related benefits to families and society are well established and accepted. Because blood is taken from essentially all newborns for NDBS in both the United States and Canada, the potential uses of this population-based pool of specimens for research and program evaluation make it almost priceless. The questions that arise relate primarily to ethics and legalities regarding specimen collection and RBS use. Because most US and Canadian NDBS systems do not adequately address education of the public (including parents), health care professionals, and policy makers on secondary specimen use, the RBS questions and controversies persist, often to the detriment of possible improvements in the public's health through research.

It is essential that primary care physicians, specialists, researchers, public health professionals, and the public are made aware of, and understand, the value of NDBS and the related research opportunities

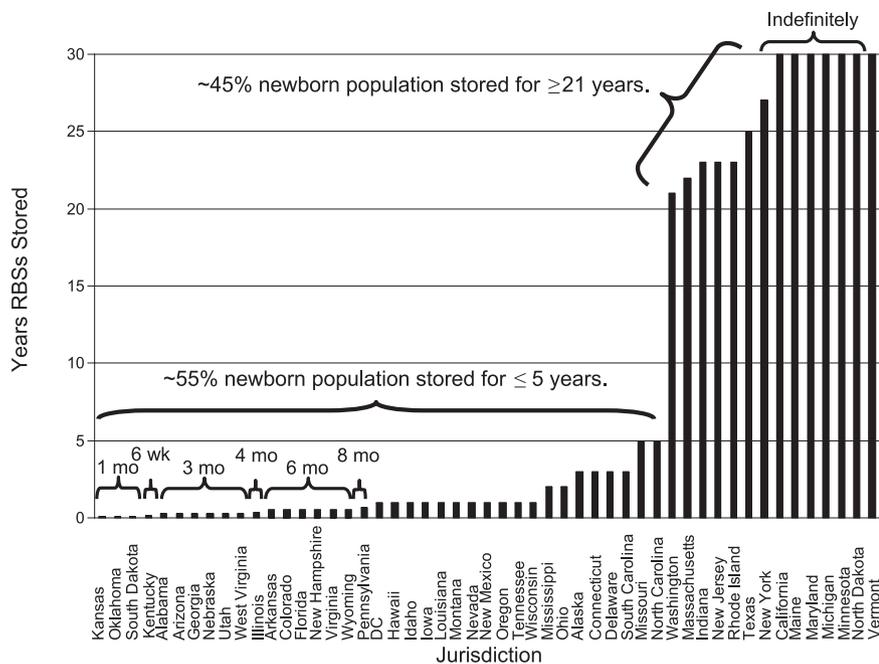


FIGURE 1

Storage time for RBSs in US newborn screening programs. Available at: <http://nnsis.uthscsa.edu/xreports.aspx?XREPORTID=18&FORMID=43&FCLR=1>. Accessed November 13, 2011. DC, District of Columbia.

that exist. Both articles in this issue of *Pediatrics* document the need for better public education and transparency in program policies, as well as the willingness of the public to support health improvement activities with RBSs once adequate information has been conveyed. Less clear is the best way in which to provide this information and the extent to which parental permission is required for secondary specimen uses. Although Botkin et al¹ concluded that US parents prefer an opt-in mechanism for specimen retention, Bombard et al² reported ambivalence among Canadian parents.

Currently, the true impact of opt-in or opt-out models for RBS storage is not known because the concepts are only now being implemented. Recent legislation in Texas has changed an opt-out option enacted by the previous legislature to an opt-in option that will begin shortly.⁵ Unfortunately, the recommendations of the Secretary of Health and Human Services' Advisory Committee on Heritable Disorders in Newborns and Children³ have not yet been acted on and therefore no studies conducted of either model. Because prenatal parent education is not yet the norm in most jurisdictions, the burden

for explaining both the NDBS program and the possible storage and uses of RBSs will primarily affect hospital nurses and primary care physicians. Confused parents will be depending on health care providers to be knowledgeable and reassuring as information about both screening and secondary potential uses of their infant's blood are discussed.

Botkin et al¹ found that the use of a movie had a significant educational impact on gaining support for the use of RBSs for secondary purposes. Although movies describing NDBS in general are common, they do not usually include references to secondary uses of RBSs. Revised materials containing additional information on specimen storage and use will be necessary additions to program educational materials in the future if transparency is to be embraced.

Clearly, the studies by Botkin et al¹ and Bombard et al² both provide useful documentation of the need for improved public and parent education. It is essential for prenatal and primary health care personnel to clearly understand their local NDBS program and secondary specimen use issues so that they can better inform families about the decisions they must make regarding NDBS and RBSs. A simple telephone call to the state health department's NDBS program⁶ should provide easy access to the necessary educational information and all program policies. Openness and transparency will resolve the public's fear of misuse.

REFERENCES

1. Botkin JR, Rothwell E, Anderson R, et al. Public attitudes regarding the use of residual newborn screening specimens for research. *Pediatrics*. 2012;129(2):231–238
2. Bombard Y, Miller FA, Hayeems RZ, et al. Citizens' values regarding research with stored samples from newborn screening in Canada. *Pediatrics*. 2012;129(2):239–247
3. Therrell BL, Jr, Hannon WH, Bailey DB Jr, et al. 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;13(7):621–624. Extended digital content available at: <http://links.lww.com/GIM/A168>. Accessed November 13, 2011
4. Watson MS, Epstein C, Howell RR, et al. Developing a national collaborative study system for rare genetic diseases. *Genet Med*. 2008;10(5):325–329
5. Becca Aaronson. Senate OKs Changes to Newborn Screening Program. *The Texas Tribune*. May 23, 2011. Available at: www.texastribune.org/texas-legislature/82nd-legislative-session/senate-oks-changes-to-newborn-screening-program/. Accessed December 15, 2011
6. National Newborn Screening and Genetics Resource Center, NBS Program Contact Information. Available at: http://genes-r-us.uthscsa.edu/State_contacts.pdf. Accessed November 20, 2011

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