Current State Practices and Policies on the Storage and Use of Newborn Screening Samples

This is a brief white paper that was commissioned by the Institute of Medicine Roundtable on Translating Genomic-Based Research for Health in preparation for its May 24, 2010 workshop on challenges and opportunities in using residual newborn screening samples for translational research. The responsibility for the content of this article rests with the author and does not necessarily represent the views of the Institute of Medicine or its committees and convening bodies.

Authored by: Alissa Johnson, MA
Principal Consultant
Johnson Policy Consulting

States screen approximately 4 million newborns annually for certain heritable disorders as set forth in state laws and policies. Upon the completion of newborn screening tests, residual dried blood specimens remain on collection cards obtained for the purpose of screening. These specimens, if stored, present a unique opportunity to advance public health research and scientific study. State practices and policies around the storage and secondary use of residual dried blood spots vary widely—from those that prohibit research use to others that declare the benefits of storage and allowable research use, if conducted in accordance with established requirements. When defined in statutes and regulations, state policies on storage and use of residual dried blood specimens address issues such as authority over the use and retention of specimens; access to specimens; permitted secondary use, if any, of specimens; and parental education about state policies on the retention and use of specimens.

All states must retain residual dried blood specimens for a brief period of time to complete the screening process, but some newborn screening programs continue to store specimens for several years or indefinitely.[1] As of May 2010, state laws or regulations in at least 18 states contain provisions that directly address the storage and use of residual dried blood specimens, and of these states only one—Mississippi—strictly prohibits the use of residual dried newborn screening specimens for research or purposes other than confirmation of test results.[2] Although the remainder of newborn screening programs likely have established positions regarding the use of residual dried blood specimens, program practices are not transparent.

[1] See the National Newborn Screening and Genetics Resource Center, National Newborn Screening Information System, for length of specimen storage by state at http://nnsis.uthscsa.edu/rde/xreports.aspx?XREPORTID=5

[2] Mississippi Dept. of Health, Chapter 38 Rules and Regulations Governing Newborn Screening, http://www.msdh.state.ms.us/msdhsite/_static/41,0,101,60.html

Authority Over the Use and Retention of Specimens

Laws and regulations in California, Maine, Utah and Washington declare that newborn screening specimens are the property of the state. Policies in several other states such as Indiana, Michigan, Missouri, Nebraska, Oklahoma and Wisconsin grant newborn screening programs authority over the retention and use of residual dried blood specimens or require the program to develop a system for handling storage and use issues. Some of these programs allow parents to determine how their samples may be used. For example, in California and Maine where researchers may have access to specimens for approved studies, a person can prohibit the use of samples for program evaluation or research through the submission of a written request. Parents also can request destruction of residual dried blood specimens after a defined period when the samples are no longer needed for screening purposes in several states, including Michigan, Minnesota, South Carolina, Texas and Washington. Most state policies that permit research use of residual dried blood specimens employ an opt-out approach, in which samples are released for approved research unless parents indicate otherwise. In Nebraska and New Hampshire, however, researchers must obtain written consent from parents of individuals whose specimens are being requested.

Access to Specimens

States with laws and regulations that allow secondary use of specimens following the newborn screening process typically limit access to health department or laboratory personnel or researchers and other individuals with departmental approval. When access to specimens is granted to researchers, policies often state clearly that the health department is responsible for preparation of specimens and other information approved for study, and, for example, in California, Missouri, Nebraska and North Dakota, the state health department may bill researchers for these services. Most states that address research use also discuss institutional review board (IRB) requirements or refer to adherence with federal regulations governing the protection of human subjects.

Some states prohibit access to personally identifiable information in conjunction with the release of specimens such as Indiana, South Carolina and Texas. In Iowa and North Dakota, state policy requires researchers to justify the need to access personally identifiable information in study proposals. Researchers who wish to access personally identifiable information associated with specimens also may need to obtain consent from parents such as in California, Maine, Texas, Utah and Washington. In California an IRB may modify the usual informed consent requirements for the release of personal information if it determines that the research has such public health value that the waiver is justifiable.[3]

[3] California Health and Safety Code §124980.

Secondary Use

While some states do not place restrictions on the types of research projects allowed, others limit secondary use of residual dried blood specimens for research purposes to specific study areas. A few states only permit research on newborn screening related issues, including Wisconsin,[4] which confines secondary use to research and evaluation purposes related to congenital and metabolic disorders or laboratory procedures, and Massachusetts, which offers participation in pilot studies of conditions that may be added to the state newborn screening panel in the future.[5] Other states permit slightly broader forms of research such as Iowa, which allows newborn screening related studies, those that would impact the health of a child from whom no other specimens are available, or studies that would inform existing public health surveillance activities.[6]

Parent Education

Policies in at least eight states require that information provided to parents regarding the newborn screening program discuss storage and use of residual dried blood specimens. Educational materials may explain options for parents, if applicable, to control specimen storage and use. In several states program websites describe approved research studies involving the use of residual dried blood specimens and findings that have brought forth advances in newborn screening as a result.

[4] Wisconsin Administrative Code, DHS 115.05.
[5] 105 Code of Massachusetts Regulations 270.
[6] 64 Iowa Administrative Code 4.3,