Storage, Retention, and Use of Residual Dried Blood Spots

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Storage, Retention, and Use of Residual Dried Blood Spots

Overview

- Storage of residual DBS by screening labs
- Retention times for residual DBSs
- Use of residual DBSs and the restrictions
- Policies impacting dried-blood spot (DBS) use
- Controversy: media, and parents
- National DBS repository: actual BDS or virtual?


“Whole blood absorbed into filter paper and then dried offers an excellent means for creating a repository (bank) of samples for DNA investigations.”

“Ideally, residual DBSs should be stored frozen (preferably at −20°C) in sealed bags with low gas permeability containing a desiccant and a humidity indicator.”
U.S. Newborn Screening Data

Comparison of Retained Residual Samples - 1993 vs. 1999 vs. 2009

Number of Programs

- Not Reporting
- Indefinitely
- 5-23 Years
- 2-4 Years
- 1 Year
- 8 Months
- 6 Months
- 4 Months
- 3 Months
- 2 Months
- 1.5 Months
- 1 Month
- < 1 Month

HTTP://GENES-R-US.UTHSCSA.EDU
Informed/Consent Issues

NOTICE OF INFORMATION PRACTICES AND PRIVACY POLICY – Effective 01/24/01

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The California Department of Health Services is authorized to collect information requested by Health and Safety Code Sections 125000, 125025, and 125030. This information is used to identify newborns with inherited or congenital disorders in order to expedite prevention or treatment of the disorder. Provision of this information is required by law (17CCR 6500 through 6510) and if not provided could result in the death or permanent handicaps for affected newborns.

Uses and Disclosures of Health Information: The California Department of Health Services uses health information about your newborn for screening, to provide health care service, to obtain payment for screening, for administrative purposes, and to evaluate the quality of care that you receive.

We may use the information and specimens obtained by participation in the program for medical research without identification of the person from which they were obtained unless you specifically request in writing they not be used by contacting the person listed below.

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我們可能把參加計劃者的資訊和樣本用於醫學研究，而不披露研究對象的身份，除非您向下文指定之人士提出特別書面請求，要求不要使用該資訊和樣本。
Some reasons for retaining residual DBSs

- Legal accountability (e.g., number of punches taken for analysis, the existence of a sample and its adequate collection)
- Future DNA testing
- Reconfirmation of newborn screening analytical results
- New method evaluations and comparisons
- Epidemiological or other public health surveys
- Special health related studies for patient or family
- Forensic studies

Some reasons for retaining residual DBSs

- Confirmatory diagnosis (reconfirm false negative or false positive finding)
- Quality assurance and public health needs (method development, epidemiological studies)
- Research uses (DNA extraction – understanding disease history; gene-environment interactions)
- Clinical testing – post mortem i.d. of disease cause
- Non-medical use – kidnapped children i.d., deceased persons i.d., paternity (subpoena), criminal i.d.

Examples of Previous Use of Residual NBS Specimens

- HIV Seroprevalence
- Diabetes type 1 risk and autoimmune disease onset
- Searching for new early markers of diseases
- Surveillance for environmental factors, infections, and genomic health issues, e.g., autism, cerebral palsy
- Determining allele frequencies for public health assessments
- Understanding hearing loss causes — CMV association
- Searching for frequency of deaths caused by SCID
- Environmental exposures: e.g., polyfluoroalkyl chemicals, perchlorate, lead
- Quality assurance — case specimen exchange among labs
National Report on Genomics and Health

Need for population-based data

- Population-based data on gene variants
  - Prevalence of gene variants
  - Association with risk of disease, death
  - Gene-environment and gene-gene interactions

- Genetic test evaluation (validity, utility)

- Development of public health interventions, e.g., newborn screening expansion

- Currently, minimal population-based data on gene variants to guide screening or interventions
Policy Statements
Residual Newborn Dried Blood Spots

AAP Task Force 2000 [Pediatrics 2000; 106 (suppl)]

- Develop policies for unlinked/linked residual samples in research/surveillance
- Organize collaborative efforts to develop minimum standards for storage of residual samples at state level
- Consider creating national or multi-state population-based specimen resource for research
Residual Newborn Screening (NBS) Specimens

A statement of position:

“There may be other reasons (other than QA) to save DBS specimens, including test development, research, and forensic identification. To retain DBSs for such purposes requires clear guidelines that are incorporated into national consensus policies that state health departments follow in carrying out their authorized NBS programs.”

http://www.aphl.org
Newborn-Blood Storage Law Stirs Fears of DNA Warehouse

By Alecia Meding

An obscure bill that sailed through Congress and was signed into law last month is striking fear of a nationwide DNA warehouse potentially open to abuse by law enforcement agencies or health insurance companies.

But proponents say the law is a much-needed mechanism to protect the genetic data produced in newborn blood screens.

The Newborn Screening Blood Law Act of 2008 (HR 6942; S 314; P.L. 110-264), signed into law on April 24, empowers a committee to provide guidelines to all states on how — and for how long — they should store blood. At present, all states store blood from newborns, and some, like California, store it indefinitely. Eight of the committee members are medical researchers, who almost universally have longer storage times, so critics fear that the national guidelines will lead to more storage of samples, which contain valuable DNA.

"What we are doing is taking an individual genetic code and saying it's the government," said Twila Brase, of the Minnesota activist group Citizens Council on Health Care. "And once we do that, it's available for whatever a legislature wants to do in 20 years. The fact of the matter is that we don't know what they could or would do.

States have been storing blood samples from newborns since blood screening for genetic defects and diseases began in the 1960s. The samples are held to detect and treat a wide range of diseases, but in the age of the genome, the issue of storing samples has taken on unprecedented importance. Blood samples contain DNA that can be anonymously linked to individuals, which may in the future present tempting data to governments, businesses and health providers.

Currently, each state has its own policy about storing newborn blood samples. California has screened and stored more than 12 million newborns' blood samples since 1980, while Texas disposed of them within months.

Brase's group wants to recall all so-called heparin destroyed.

"You're building an entire DNA warehouse for the public without the public's consent," Brase said. "Who will own the DNA of the citizens and what is that going to mean? And what we're doing is pushing an entire genetic research program on the population without the consent of the population."

Proponents, however, say the scientific and medical value of the blood samples far outweigh the privacy risks of storing biological materials from every newborn.

"They are extremely valuable when they are anonymized for research when looking at new technologies," said Edward Howell, chairman of the committee referred to in the bill, the National Councils on Birth Defects and Safety. "Renes in newborns and adults can be used in the Health Resources and Services Administration. These consensus theories are very popular on the blogs, but...the states have been very careful in dealing with blood samples.

Howell says law enforcement agencies have asked states for blood samples and been turned down.

"The bottom line is that many states have kept them for a very long time and I am unaware of anything that has been done with them that would concern even a very conservative person," Howell said.

Ted McCoy, co-director of the VCLA Center for Society and Genetics and co-author of DNA Privacy and Policy, agreed that the states have been trustworthy guardians of their biobanks.

McCoy said that even in the case of the identification of a missing child, California's health regulations still turned down a law enforcement request to use a blood spot.

He applauded that decision and referred to it as a clear policy for dealing with the samples and limiting their use.

"We actually think they ought to be a firewall between forensic and medical uses," McCoy said. And he argued that — from a health care perspective — the samples are extremely and uniquely valuable.

"It's one of the most unbiased sets of samples of the newborn population of a state," McCoy said.

Donna Lerner, who served as counsel for the Massachusetts Department of Public Health and the New York State Senate, said that the bill's policy had the right ethical safeguards in place.

"Groups are told prior to screening that the racial component is kept for at least 10 years," she wrote in a e-mail.

"She also noted that all research conducted on residual specimens has to be approved by an Institutional Review Board, which is an ethical guidelines for human experiments. The study also requires that research on identified specimens only be conducted with the informed consent of the subject, or a parent guardian."

These are the types of checks-and-balances that the newly empowered committee could take up during their next meeting this September, when they will begin drafting the state guidelines.

One thing is for sure: As scientists and law enforcement officials continue to learn more about how to use the DNA in each of our cells, the way ancestral samples are handled by the government seems certain to receive more scrutiny.

"The whole confidentially issue is certainly a huge issue in the age of genomics," McCoy said.
“Additionally, storage and secondary uses have been documented to occur without parental consent.”

“In the absence of uniform guidelines there is an urgent need to develop policies that address the issues of DBS storage and their secondary uses, and the ensuing ethical, legal, and social dilemmas.”

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hough in existence for over thirty-five years, due to the increasing panoply of possible tests, newborn screening programs are drawing public attention. Many jurisdictions have mandatory newborn screening programs for treatable disorders. Disorders are detected through tests on blood spots drawn from a newborn’s heel soon after birth and verified through a diagnostic test with follow-up. Unbeknownst to most parents, these blood spot cards are also stored thereafter. Indeed, while dried blood spots (DBSs) are primarily used for screening for health problems, experience demonstrates that they can be made useful in various contexts unrelated to screening.

Newborn dried blood spots have taken on a new life as a result of developments in genetics and the increasing ability of new technologies to link DNA information with clinical data. Additionally, storage and secondary uses have been documented to occur without parental consent. In the absence of uniform guidelines, there is an urgent need to develop policies that address the issue of dried blood spot storage, secondary use and the ensuing ethical, legal, and social dilemmas.

Internationally, regionally, and nationally, government, professional, and consumer organizations have contributed to the debate on the storage and retention of newborn screening residual blood samples. Despite all these efforts, a consensus of opinion on any one issue has yet to be reached. We will compare current guidelines and policy documents that apply to banking DBSs and assess the similarities and differences as concerns consent to storage, length of storage, and access to stored samples. Our comparison examines countries from different regions of the world and offers different socio-political contexts for examining the rationale for storage and issues of confidentiality and consent. As novel uses of newborn spots emerge, and as researchers and public officials contemplate mechanisms for the retention of DBSs by newborn screening laboratories, it is crucial to outline current purposes and lengths of storage and adequate consent requirements for the secondary uses of archived blood spots in research or otherwise.

**Banking Residual DBSs: Purpose and Length?**

**Purpose of Storing**

Since the late 1960s, newborn screening to detect congenital metabolic disorders has been standard pediatric procedure in newborn care in most industrialized countries. Early detection of pre-symptomatic disorders such as Phenylketonuria (PKU) and Congenital hypothyroidism (CH) has prevented chil-

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Texans unknowingly donate children's blood to research

Medical privacy advocates, ethicists say parents should be asked for consent before newborns' screening samples are kept.

By Mary Ann Roser
AMERICAN-STATESMAN STAFF

Sunday, February 22, 2009

For almost seven years, the state has been indefinitely storing blood from nearly all newborns in Texas without their parents' consent for possible use in medical research.

The blood is collected as part of a 44-year-old state-mandated newborn screening program in which hospitals, birthing centers and midwives draw blood from a baby's heel — parental consent isn't required for that, either — so the state can test for a host of birth defects. The state either discarded the blood after six months or, more recently, stored it for three years before destroying it.

But starting in 2002, the state health department began collecting and keeping blood indefinitely for current or future medical research, a practice that has been the subject of a legal challenge in Minnesota.

Five dots of blood are collected on paper for the screening and then stored.

Under the health department's policy, the samples can be used by the medical community for things like cancer research, birth defects studies and calibration of lab equipment, said Doug McBride, spokesman for the Department of State Health Services.

“... without the parents’ consent for possible use in medical research.”
Purpose: To Develop a Strategic Plan to Assess the Feasibility, Utility, and Practical Implementation of Establishing a National/Multi-state Bank of Residual Newborn DBS

Host: Mary Lou Lindegren, MD
Centers for Disease Control and Prevention
Objectives of the Meeting

- Outline potential uses of banks for public health
- Review experiences using state-based spot banks for public health applications
- Assess storage, laboratory, and database issues
- Propose multi-state models for the future
- Review feasibility issues-challenges + barriers
- Update status of state storage and use policies for leftover specimens
- Design strategic plan for banking implementation
Summary of State Policy Data – CDC 2003

- State-to-state variability in residual blood spot storage duration and adherence to suggested storage guidelines

- 45% of states had written guidelines concerning the uses of their residual samples

- 16% informed parents DBSs retained

- Nearly 80% of states favored future storage of identifiable samples at state level

Assuming funding is available, in which type of facility would you prefer to store residual NBS specimens? (2000 data)
Challenges

- Resources
- Data sharing issues
- Confidentiality, security, privacy issues
- IRB (ethical reviews)
- Legal, ethical, social issues
- Informed consent issues
- Education efforts for parents and others
- Maintain primary functions of NBS program
Outcome – Develop a Strategic Plan for a Virtual Database of Available Specimens for Research Use

- Create a working group to develop and publish a strategic plan for implementation
- Establish a central gatekeeper
- Establish criteria for inclusion, access and use
- Develop consensus standards for storage, QA, and cataloging/retrieval, data elements, linkages
- Plan Pilot Studies to demonstrate usefulness
- Address gaps + feasibility issues
- Larger stakeholders meeting for buy in
Current Thinking

- Still need to develop state policies on retention, storage, and use.
- NIH funding long-term outcome database for rare conditions diagnosed through newborn screening.
- Virtual specimen database for use in conjunction with the long-term outcomes database is possible.
- States appear interested in collaborating.
- Tendency towards referring to residual spots as patient “record” for policy implementation.
Thank You!!