Storage, Retention, and Use of Residual Dried Blood Spots

Discussion Webinars

Brad Therrell, PhD and Harry Hannon, PhD

University of Texas Health Science Center at San Antonio
and
Newborn Screening and Genetics Resource Center in Austin, Texas
Draft ‘White’ Paper for ACHDNC Working Group

Workgroup Chairs:
Brad Therrell, Ph.D
Harry Hannon, Ph.D

Workgroup Members:
Don Bailey, Ph.D
Alan Fleischman, MD
Ed Goldman, JD
Jana Monaco
Bent Norgaard-Pedersen, M.D., D.MSc.
Sharon Terry, MA

HRSA Staff:
Alaina Harris, MSW, MPH
Purpose of Webinar

To provide the newborn screening stakeholder community with information about a subject of special interest to the ACHDNC and to solicit outside input into the preparation of a discussion ‘white paper’ that may lead to further ACHDNC actions.
Storage, Retention, and Use of Residual Dried Blood Spots

Overview of the Issues

- Storage of residual DBS by screening labs
- Retention times for residual DBSs
- Uses of residual DBSs (and the restrictions)
- Policies impacting dried-blood spot (DBS) use (privacy issues)
- Public relations: media, parents (privacy issues)
- National (multi-state?) DBS repository: actual or virtual?
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


These guidelines provide scientific information for policy development by state health departments considering appropriate use of newborn screening specimens after screening tests are finished. Information was collected, debated, and formulated into a policy statement by the Newborn Screening Committee of the Council of Regional Networks for Genetic Services (CORN), a federally funded national consortium of representatives from 10 regional genetics networks. Newborn screening programs vary widely in approaches and policies concerning residual dried blood spot samples (DBS) collected for newborn screening. Recognition of the epidemiological utility of DBS samples for HIV seroprevalence surveys and a growing interest in DBSs for DNA analysis has intensified consideration of issues regarding retention, storage, and use of residual DBS samples. Potentially these samples provide a genetic material "bank" for all newborns nationwide. Their value as a resource for other uses has already been recognized by scientists, administrators, and judicial officials. Programs should formulate rules for retention and use of residual newborn screening DBS samples based on scientifically valid information. Banking of newborn samples as sources of genetic material should be considered in light of potential benefit or harm to society.

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BACKGROUND

The Council of Regional Networks for Genetic Services (CORN) is a federally funded project to improve the quantity, quality, and availability of cost-effective genetic services in the United States. CORN was developed in 1985 in response to the need for an organization that could coordinate activities among federally funded genetic service networks encompassing the entire United States and could implement programs of national significance that emerge from regional initiatives in priority areas such as quality assurance, data collection, and education. Two delegates from each of the 10 defined networks serve on the CORN steering committee with additional representation from the Alliance for Genetic Support Groups, national sickle cell disease programs, and certain other organizations involved in genetic services. CORN members constitute a unique organization of genetic service providers, public health personnel, and consumers. In its goals
Some reasons for retaining residual DBSs:

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

“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.”

Thorough 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 spots are also stored afterward. 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 bioinformatics 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, governmental, 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 newborns 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 bloodspots 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 paediatric 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-
Danish Biobank

Established in 1993
Institutionalized in 2004

“A biobank is … a structured collection of human biological material which is accessible under certain criteria, and where information contained in the biological material can be traced back to individuals.”

Bent Norgaard-Pedersen
Statens Serum Institut
Copenhagen, Denmark
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.”

Source: http://www.aphl.org
Residual Newborn Screening (NBS) Specimens

A statement of position:

- Residual Dried blood spots are a valuable national resource that can contribute significantly to the health of children.
- NBS blood spots are stored with rigorous control and respect for privacy and confidentiality.
- Parents should have the option to have their child’s specimen stored in a national repository for research.”
Reported Residual Bloodspot Storage – 5/1/2009
(Ascending Order)

~ 54% Newborn Pop. Stored for ≥18 yrs.

~ 46% Newborn Pop. Stored for ≤3 yrs.

Years Residual Dried Bloodspots Stored

Kansas
Louisiana
Oklahoma
South Dakota
Georgia
Alabama
Arizona
Nebraska
West Virginia
Georgia
Alabama
Arizona
West Virginia
Illinois
Arkansas
Connecticut
Kentucky
Missouri
Nevada
Washington
Pennsylvania
Hawaii
Idaho
Nevada
New Mexico
Oregon
Tennessee
Wisconsin
Dist Columbia
Mississippi
Ohio
Utah
Alaska
Montana
South Carolina
Massachusetts
Indiana
New Jersey
Rhode Island
Texas
New York
Florida
Iowa
Maine
Maryland
Michigan
Minnesota
North Carolina
North Dakota
Vermont

Program Location

Indefinitely

1 mo
3 mo
6 mo
6 wk
4 mo
Considerations and Recommendations for a National Policy Regarding the Retention and Use of Dried-Blood Spot Specimens after Newborn Screening

INTRODUCTION
BACKGROUND
LEGAL AND ETHICAL ISSUES
POLICY ISSUES
SCIENTIFIC ISSUES
FINANCIAL ISSUES
CONCLUSIONS/RECOMMENDATIONS
REFERENCES
Recommendation 1

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.
Recommendation 2

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.
Recommendation 3

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.
4) All state newborn screening programs should work proactively to ensure that all families receiving prenatal care are educated about newborn screening.
Recommendation 5

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).
Recommendation 6

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.
Recommendation 7

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

a) Model consent/dissent processes on the use of residual specimens in newborn screening;

b) 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;
Recommendation 7
(Continued)

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

c) National data on the utility of any additional consent/dissent processes implemented relative to potential research uses of residual newborn screening specimens;

d) Educational materials with facts about potential uses of residual newborn screening specimens for both consumers and prenatal healthcare providers.
You may email comments to:

therrell@uthscsa.edu

or

hhannon@cdc.gov