

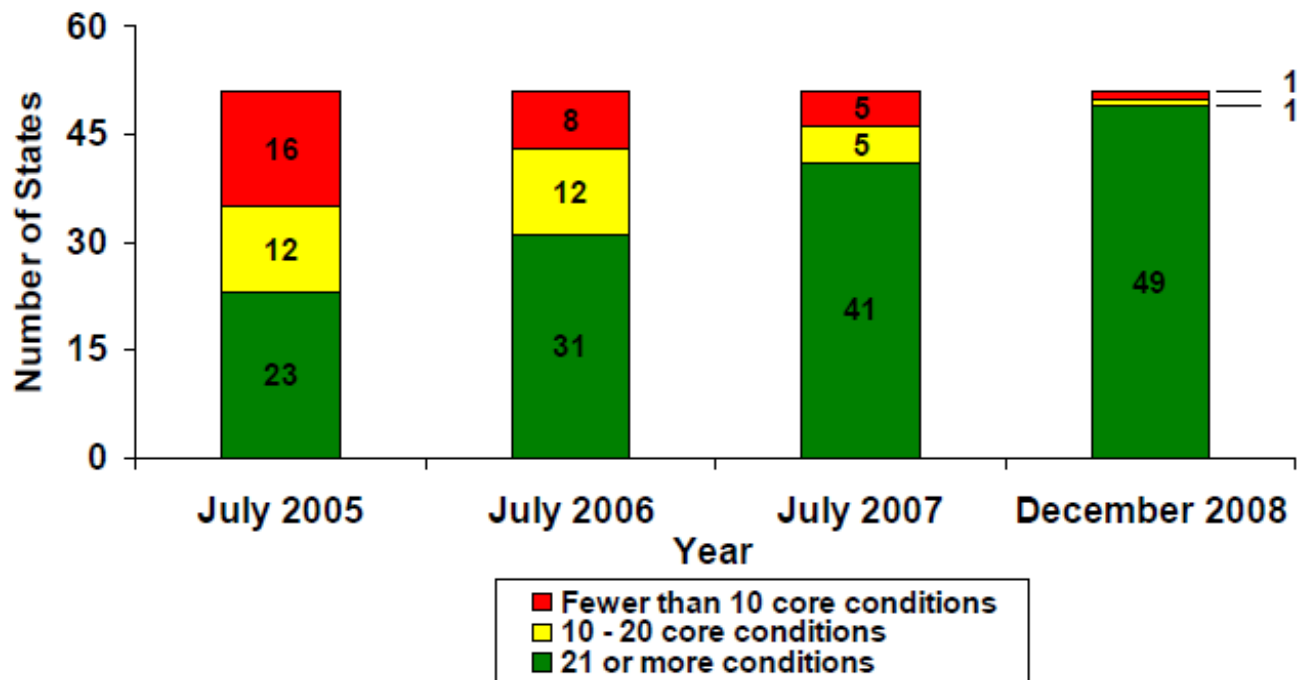
Newborn Screening Ethics and Policy

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History

- **1960' s: Dr. Robert Guthrie- Newborn screening programs began in the US**
 - Phenylketonuria- using blood collected on filter paper from a baby's heel
- **1970' s: Congenital hypothyroidism was the next disease widely added**
- **1990' s: Tandem Mass spectrometry was developed and was piloted in some NBS programs**
- **2000 : Statement was released by ACMG/ASHG recommending MS/MS be thoughtfully integrated into NBS programs**
- **2004: U.S. Secretary of Health and Human Services establishes a federal 'Advisory Committee on Heritable Disorders in Newborns and Children' (ACHDNC)→ SACHDNC→ DACHDNC**
- **2005: ACHDNC recommended that the number of disorders states were screening for be expanded from 9-29 (due to MS/MS technology)**
- **2008: The Newborn Screening Saves Lives Act**
- **As of April 2013 the DACHDNC- 57 Conditions: Recommended Uniform Screening Panel**

Newborn Screening Tests



Source: March of Dimes. Data reported from NNSGRC.

What's on the Panel?

- The panel differs from state to state
- Most states have mandatory NBS with defined opt-out policies
- Adding tests: complex social, ethical, and political issues
- Technological Advancements → increased ability to add tests. **Just because we CAN, does not mean we SHOULD**

Adding New Conditions

- Main Considerations:
 - The condition
 - Onset
 - Severity
 - Disease Course
 - Testing
 - Technical and Financial Feasibility
 - Treatment
 - Is Treatment Available?
 - Is Treatment Effective?
 - Other
 - The Diagnostic Odyssey

ACHDNC Evidence Review Process: Overview

State Specific:

Emma's Law: NJ- Lysosomal Storage Disorders

- Krabbe
- Pompe
- Gaucher
- Niemann- Pick
- Fabry

Brian's Hope: CT- Adrenoleukodystrophy

Dried Blood Spots

Why We Store Dried Bloodspots

- **Newborn Screening Program Operations:**
 - Verification of testing results
 - Quality control and quality assurance
 - Development of improved methodologies to reduce false positive and false negative NBS results
- **Public Health Related:**
 - Development of new tests to screen for more disorders
 - Public health studies to better understand conditions in the general population for the benefit of all.
 - Basic scientific studies to better understand the causes of birth defects, cancer, or chronic disease
- **Other Uses**
 - Testing to assist in identifying a missing or deceased child
 - Testing, as requested by the family, to provide additional medical information

Reported Residual Bloodspot Storage – 5/1/2009

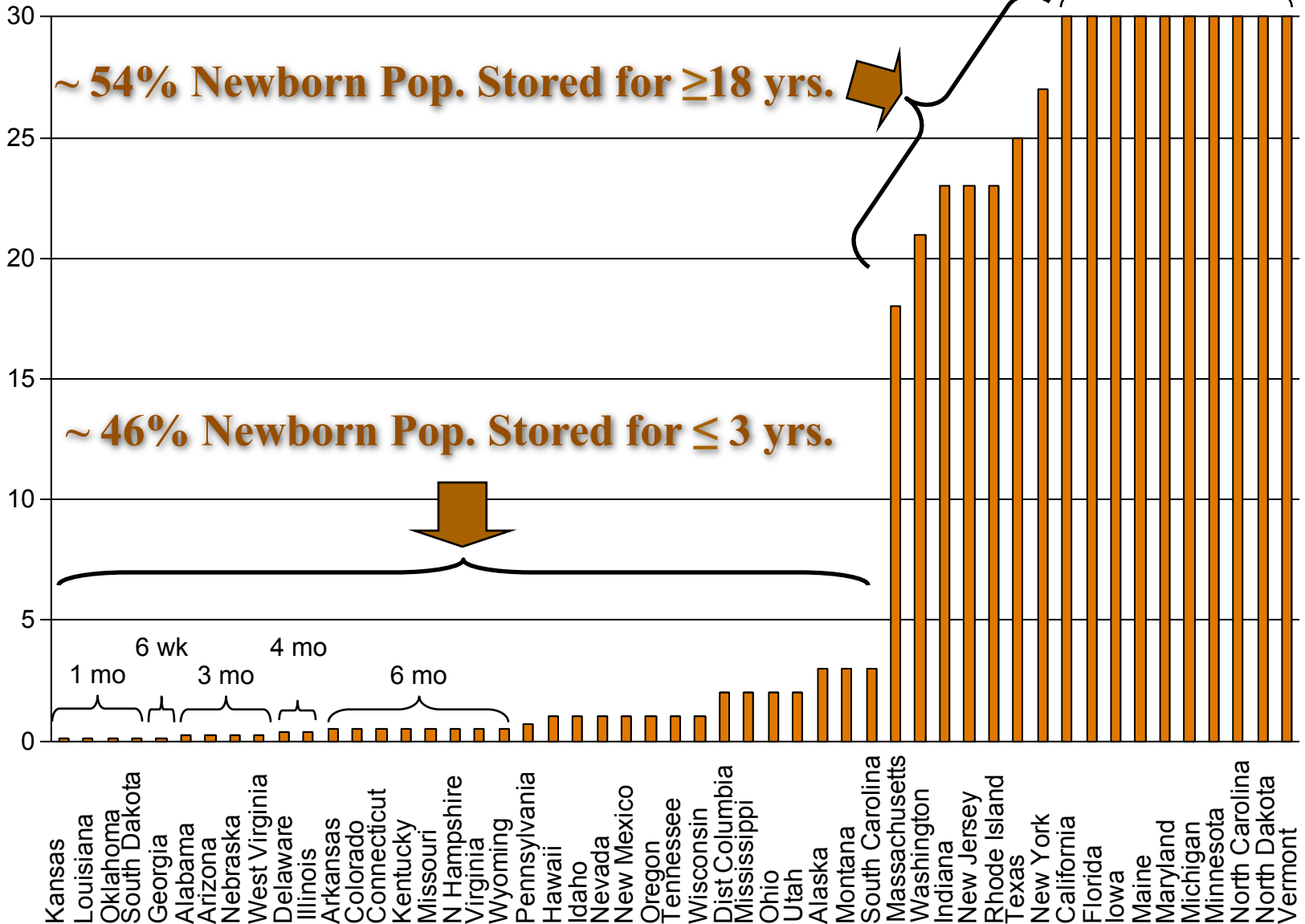
(Ascending Order)

Years Residual Dried Bloodspots Stored

Indefinitely

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

~ 46% Newborn Pop. Stored for ≤ 3 yrs.



Program Location

What Does The Public Think?



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Newborn Blood Spot Litigation: 70 Days to Destroy 5+ Million Samples

Posted by [Adam Doerr](#) on February 2, 2010



Sometime in the next few months, Texas will destroy more than 5 million blood samples collected from newborn babies across the state over the past seven years. The lawsuit that led to this result—agreed to as part of a [settlement](#) reached between the state and a civil rights group representing a group of parents—illustrates a number of interesting points about the law and litigation of genetics issues.

As we discussed in [A Closer Look at Biobanking of Newborn Blood Spots](#), states collect blood samples from most infants born in the United States each year, with the goal of detecting and treating a variety of potentially serious conditions. The [Texas Department of State Health Services](#) (DSHS) has been collecting newborn blood samples from babies born within the state since the 1960s. Texas currently tests for conditions

including cystic fibrosis, endocrine disorders, fatty acid disorders, and others—[28 disorders in all](#) (pdf). At least some of the samples are apparently subjected to genetic testing for [hemoglobinopathy](#), [phenylketonuria](#), and [galactosemia](#).

What Does The Public Think?

CNN: February 4, 2010



Legal Issues In Texas and Minnesota

<http://www.cnn.com/2010/HEALTH/02/04/baby.dna.government/index.html>

Informed Consent: A Perfect World!

- Most states DO NOT require Informed consent for NBS:
 - Individuals who collect NBS samples often lack knowledge regarding the concept of false positives and familiarity with the conditions (Lowenstein [2010](#); Tluczek et al. [2009](#)).
 - No guidelines: how to ensure that parents understand NBS' role in disease detection.
 - Parents often not informed about Blood Spot Storage.
- 1996 – CORN Newborn Screening Committee:
“Appropriate consent is an important issue. Some legal experts have proposed that proper consent is impossible since it is not possible to adequately inform or educate parents about all potential uses....”

Informed Consent In Research

- Institutional Review Boards (IRB' s) review
- Federal regulations require the inclusion of both risk and benefit information in the informed consent process
- Many Things To Address:
 - Potential future research use of samples
 - Release of data to a public database
 - Risks, benefits and limitations of the study
 - Release of incidental findings (IF)
 - Return of Individual Research Results (IRR)

What Do Parents Want?

- **Bombard et al. 2012: Citizens' Values Regarding Research With Stored Samples From Newborn Screening In Canada**
 - 8 Focus Groups
 - Support Storage:
 - Quality control (98%)
 - Confirmatory diagnosis (98%)
 - Future anonymous research (91%)
 - Agreed that they should be able to **choose without pressure** if they want their infants dried blood stored (77%)
 - Agreed that parents should be strongly encouraged to have their infants dried blood spot stored (77%)

What Do Parents Want?

- **Botkin et al. 2012: Public Attitudes Regarding the Use of Residual Newborn Screening Specimens for Research**
 - Focus Groups, Telephone Surveys, Internet Surveys
 - 55% of Respondents were aware that NBS was done, but knowledge was limited
 - Most- Very (65.4%) or Somewhat (29%) supportive of NBS
 - Is it ok to do NBS without permission?
 - 25.5%- Definitely alright; 25% Alright; 18% Probably alright; 32% definitely not alright
 - Blood spot storage
 - 25% Not concerned, 30% Very concerned
 - 80% said definitely or probably alright to use spots for: Quality control, Maternal/child research/general public research
 - 62% Said samples should ONLY be kept if parents signed a form.

What Do Parents Want?

- **Tarini et al. 2009: Not without my permission: Parents Willingness to Permit use of NBS Samples for Research**
 - Internet survey
 - Use of NBS blood spot research
 - If Permission obtained 76.2% very, somewhat supportive
 - If Permission is Not obtained 28.2% very or somewhat supportive

The Mantra of Genetic Counseling: *Nondirective*

- Educated decisions
- Respect for patients beliefs and values
- Informed consent
- Support

Newborn Screening: Education, Consent, and the Residual Blood Spot. The Position of the National Society of Genetic Counselors

- **Newborn Screening**

The National Society of Genetic Counselors (NSGC) **strongly supports newborn screening for the uniform screening panel of conditions recommended by the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children.** When considering screening, parents should be informed of the significant and potentially fatal childhood health risks that these conditions cause.

NSGC strongly recommends that parents receive newborn screening **education and resources from a qualified healthcare provider** during the prenatal and the immediate postnatal period. NSGC supports retaining newborn screening results for a minimum of 2 years, in compliance with Federal regulations (Adopted February 18, 2012)

Newborn Screening: Education, Consent, and the Residual Blood Spot. The Position of the National Society of Genetic Counselors

- **Blood Spot Storage and Use**

The National Society of Genetic Counselors **supports storage and use** of blood spot samples by newborn screening laboratories and **transparent policies** that govern these activities. Dried blood spots are valuable resources that significantly contribute to lifesaving newborn screening and advancing public health. They are critical for program quality assurance, new test development and **institutional review board-approved research**. As such, parents should be fully informed of their options through **comprehensive education** during the prenatal and immediate postnatal period regarding blood spot storage and use policies (Adopted February 18, 2012).