Translational Research in Newborn Screening: Development of a Virtual Repository of Residual Dried Blood Spots

Amy Hoffman, MPH
Newborn Screening Translational Research Network (NBSTRN)
Outline

- The NBSTRN
- The VRDBS
- Partnering with Stakeholders
- Protecting Privacy
- Information Security
- Functional Assessment

* No financial disclosures
The Newborn Screening Translational Research Network (NBSTRN)

- Newborn Screening Saves Lives Act of 2007
- Hunter Kelly Newborn Screening Research Program
- 5-year contract from NICHD to ACMG
- Develop a research infrastructure to support investigators with projects related to newborn screening
NBSTRN Scope of Work

Networks
- Clinical specialists
- State NBS laboratories & programs

Informatics
- Establish and administer an NBS biospecimen repository
- Facilitate research on development of new methods and technologies

Facilitate Research
- Natural history studies including outcomes
- Novel screening technologies
- Novel therapies
- Genomics

Focus
- Ethical, legal and social issues
- Study planning
- Data aggregation and discovery
- Statistics
Region 4 Stork (R4S)
- Analytical and clinical validation
- Laboratory protocols, definitions

Longitudinal Pediatric Data Resource (LPDR)
- Secure, consensus-driven, standards-based clinical data collection & management
- Aggregate, share, and analyze data

Virtual Repository of Dried Blood Spots (VRDBS)
- Search & request de-identified residual dried blood spots
- Secure, research support, & request management
Virtual Repository of Dried Blood Spots (VRDBS)

- **Secure, centralized & web-based**
- **Pilot phase 6/12 to 9/12 – production date: 9/26/12**
- **Inventory of DBS samples – over 2.6 million**
- **Investigators** can request letters of support, submit questions to participating states, browse & request specimens, track shipments & provide feedback
- **States** can respond to questions, review & manage requests, approve requests & control distribution
VRDBS Overview

Investigators search for and request DBS specimens.

State programs upload de-identified data on a regular basis.

Investigators conduct research and return remaining specimen as required.

State programs review requests and ship specimens directly to investigator.

VRDBS SECURITY

- Is web-based
- Is 508 and FISMA compliant
- Uses secure user ID/password
- Uses role-based security administration
- Includes session timeout
- Maintains audit trails

Newborn Screening Translational Research Network
20 investigators have registered (1 more pending)
2 requests for specimens approved
2 questions submitted and responded to

* As of April 24, 2013
Engaged Stakeholders in 5 major ways:

- Rules under which the VRDBS operates
- Privacy policies and site user agreements
- Data Sharing Agreements with state programs
- Material Transfer Agreements
- Functional Assessments
Protecting Privacy

External ID  State  Birth year  
Storage Condition

Birth weight  Gender  Age at Collection  
Race

Ethnicity  Condition  Nutritional/Feeding Status  
Status
## Search Results

♦ **You specified:**

- **Diagnosed Condition:** Primary Congenital Hypothyroidism - CH
- **Birth Year (yyyy):** 2011 - 2013
- **Ethnicity:** Hispanic, Latino/a, or Spanish origin
- **Sex:** Female

⚠️ The NBSTRN policy is to not list the number of specimens from a state if there are less than five. Your search resulted in less than 5 specimens that are from the following state(s):

- **Michigan**

Please broaden your search (i.e., increase the year range and/or limit other search criteria). If this is still not resolved, please contact the state directly by clicking the state name above.

<table>
<thead>
<tr>
<th>State</th>
<th>Stored Specimens</th>
<th>Qty Requested</th>
<th>Usage Restrictions</th>
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<tr>
<td>California</td>
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<td>DBS Specimen Use Restrictions</td>
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<tr>
<td>New York</td>
<td>12</td>
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<td>DBS Specimen Use Restrictions</td>
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1-2 of 2 Results 1 of 1 Page

These numbers reflect only those cases in the virtual repository, not the incidence of the condition in the population.
Information Security

- De-Identified data
- Registration approval
- Secure user ID and password
- Session timeout
- Role based security administration
- Secure data transfer for data imports
- Participating states retain control and distribution of their DBS
• Federal Information Security Management Act (FISMA)
  • Standards for categorizing information and information systems
  • Guidance for selecting appropriate security controls for information systems
  • Guidance for monitoring the security controls and the security authorization of information systems
• Categorization of Low, Moderate or High risk
• Annual review of system
Functional Assessment

- Homepage revamped
- Privacy policy link on homepage
- Account registration process streamlined
- Search page updates
- Developed research support section
- 6 simple steps for “how to use this site”

Scenario 1
Reporter tries to register for an account
Denied
Investigator tries to register for an account
Approved

Scenario 2
Investigator searches for 50 condition-specific specimens
Investigator searches for 200 general population specimens

Scenario 3
Investigator adds missing information to saved draft
Completes request process

Scenario 4
State administrator reviews requests
Approves or denies
Investigator receives notification

Scenario 5
State administrator adds personnel as users of the system
State administrator modifies state information

Scenario 6
State lab technician edits specimen information
State lab technician removes specimen from system
Coming to Consensus

VRDBS

NBSTRN Workgroup Members

Functional Assessment Volunteers

State NBS Programs and Stakeholders

Newborn Screening Translational Research Network
# Acknowledgements

## NBSTRN Team

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<thead>
<tr>
<th>Name</th>
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<td>Barry Thompson</td>
<td>Meredith Weaver</td>
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## 5AM Solutions Team

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<tr>
<td>Erica Marrari</td>
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## NBSTRN Workgroup Members & NIH Investigators

## Functional Assessment Volunteers

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<tr>
<td>Rebecca Anderson</td>
<td>Natasha Bonhomme</td>
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<td>Alfredo Tirado-Ramos</td>
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Newborn Screening Translational Research Network
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<tr>
<td>Robin Cooley</td>
<td>Stan Berberich</td>
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