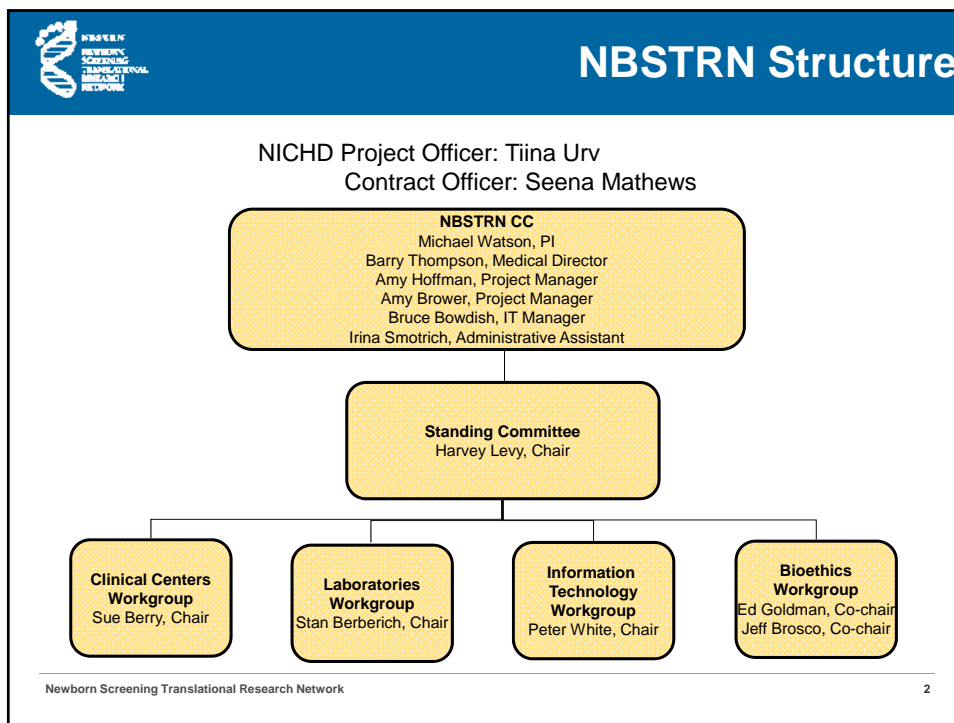



The slide features a vertical stack of four colored boxes on the left: yellow (with the American College of Medical Genetics logo), red, blue, and green (with the NBSTRN logo). The main content area has a blue-to-white gradient background.

NBSTRN Update

Amy Brower, PhD
Project Manager






NBSTRN Target Audience

- ◆ **Newborn Screening Researchers** ★
- ◆ **HRSA Genetics Collaboratives**
- ◆ **State Laboratories**
- ◆ **Clinicians**
- ◆ **Families/Consumers/Patients**
- ◆ **Rare Disease Consortia**
- ◆ **Advocacy Groups**
- ◆ **Policy Makers**
- ◆ **Industry**

Newborn Screening Translational Research Network

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NBSTRN Resources & Services

- ◆ **Residual Dried Blood Spots**
- ◆ **LTFU Data**
- ◆ **IRB Assistance**
- ◆ **Contacts/Networking**
- ◆ **Navigation**
- ◆ **IT Infrastructure**
- ◆ **Statistical Consulting**
- ◆ **Study Design**
- ◆ **Regulatory Guidance**
- ◆ **State/Federal Information**
- ◆ **Practice Guidelines**
- ◆ **Procedure Development**
- ◆ **Clinical/Natural History**
- ◆ **Money – not direct funding for research**

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Current Projects

During Year 1-2:

- ◆ **Established Standing Committee & Workgroups**
- ◆ **Defining minimum data set for LTFU**
- ◆ **Collected NBS laboratory information**
- ◆ **Website development and launch**
- ◆ **Considered IT Infrastructure options and designs**
- ◆ **Develop collaboration with APHL & Genetic Alliance**
- ◆ **SCID and LSD Pilot Projects**
- ◆ **Subcontract with 5AM Solutions, Inc. for Virtual Repository of DBS**
- ◆ **Subcontract with Mayo to adapt R4S database**

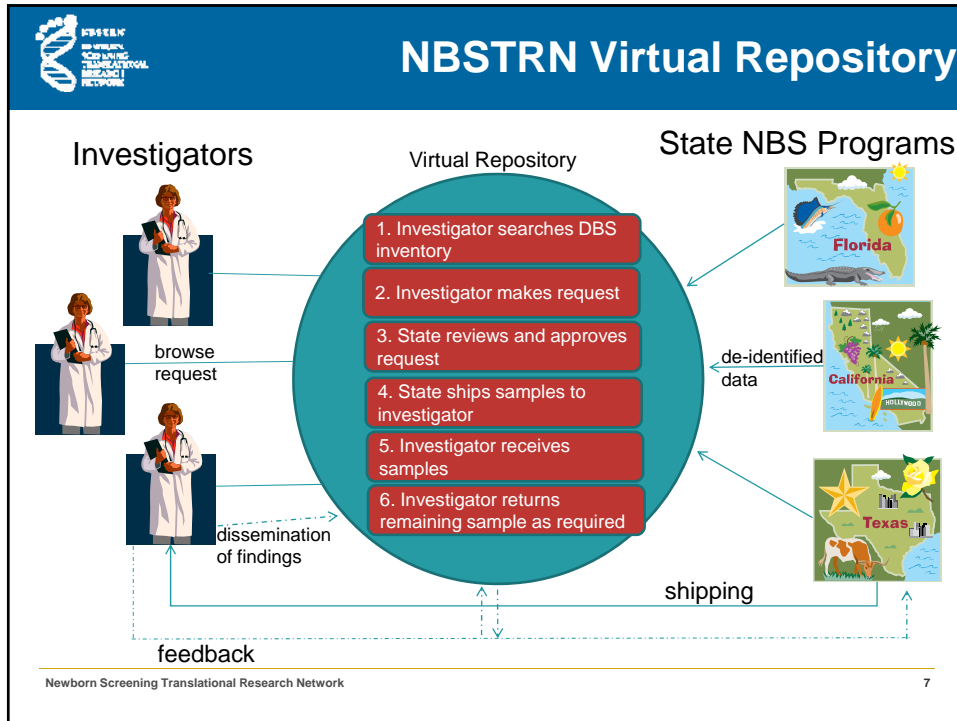
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Virtual Repository Overview

- ◆ **Centralized**
- ◆ **Web-based**
- ◆ **Inventory of available DBS samples**
- ◆ **Allows Investigators to browse and query**
- ◆ **Tools to review and manage requests**
- ◆ **Approve requests**
- ◆ **Track shipments**
- ◆ **Provide feedback**

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-
- The slide is titled 'Repository Information' and lists the data points for each available sample. The NBSTRN logo is in the top left, and the page number '8' is in the bottom right.
- ◆ **For each available sample:**
 - **State**
 - **Birth year**
 - **Gender**
 - **Birth weight**
 - **Ethnicity (Hispanic/Non-Hispanic)**
 - **Race (NIH categories)**
 - **Screening Result (screen negative or true positive)**
 - **Storage Condition (Freezer, refrigerator, room temperature)**

NBSTRN Dried Blood Spot Virtual Repository

NBSTRN
NEWBORN SCREENING TRANSNATIONAL RESEARCH NETWORK

Request Dried Blood Spots for Research
Search and request anonymized residual dried blood spots to use in qualified newborn screening related research projects.

Learn More Register Now

Sign In
Email Address
Password
Sign In
Forgot password? | Register

Browse By Participating State
- Select a U.S. State -
Why is my state not listed?
States that participate

Browse by Condition
Acetemia (disorder) (1)
Acyl-CoA dehydrogenase deficiency (disorder) (1)
Arginase deficiency (disorder) (2)
Argininosuccinate lyase deficiency (disorder) (1)
Carnitine acylcarnitine transferase deficiency (disorder) (1)
Carnitine deficiency (disorder) (1)
Carnitine palmitoyltransferase I deficiency (disorder)

Welcome
The NBSTRN Dried Blood Spot Virtual Repository is a centralized, web-based source for access to samples for newborn screening related research. This virtual repository links information from participating states and provides a de-identified view of stored dried blood spots, allowing researchers to browse and query for samples. For public health departments, the virtual repository provides the ability to review and manage sample requests, configure approved orders, track shipments, and view researcher responses to the samples they have received.
Learn More | Register Now

Done Internet 100%


R4S Database Adaptation

NEWBORN SCREENING COLLABORATIVE PROJECTS

Welcome to the NBS Portal

MS/MS SCID LSD


Done Internet 100%



Future Activities

- ◆ **Characterize NBS labs**
 - **capacity, technical capability, policies**
- ◆ **With 5AM Solutions – refine Virtual Repository**
- ◆ **Identify Virtual Repository participants and needs**
- ◆ **Refine R4S Database for LSD & SCID**
- ◆ **Develop LTFU data dictionaries**
- ◆ **Standardize LTFU language**
- ◆ **Develop bedside data capture tool and centralized database for LTFU data**


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Future Activities (2)

- ◆ **Clinical groups for pilot studies**
- ◆ **Patient specimen and re-contact repositories**
- ◆ **Refine website**
 - **Secure portal**
 - **FISMA compliant**
- ◆ **Define bioethical issues and develop policies and procedures**
- ◆ **Identify funding opportunities**
- ◆ **Plan continuation of network beyond contract**

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Acknowledgement

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