

Where can you see us next?

Consent Consensus Meeting  
September 15-16, 2011  
Salt Lake City, UT

ASHG Annual Meeting  
October 11-15th, 2011  
Montreal, Canada

APHA Annual Meeting  
October 29 –November 2nd, 2011  
Washington, DC

APHL NBS & Genetic Testing Symposium  
November 7-11th, 2011  
San Diego, CA

Do you know of a good venue for a presentation from the NBSTRN?

CONTACT US!

NBSTRN

Highlights!

Check out ALL of the postings on your workgroup WIKI!

Scheduled ONE more functional assessment with researchers on September 19th!

Preparing for the VRDBS Pilot with real data, states, and investigators in January 2012.

# NBSTRN NEWSLETTER



NBSTRN  
NEWBORN  
SCREENING  
TRANSLATIONAL  
RESEARCH  
NETWORK

September 2011

## Welcome

Welcome to the second issue of the NBSTRN Newsletter! We hope you find the Newsletters useful and informative. Please feel free to provide feedback, ideas, questions regarding this newsletter at anytime by emailing the NBSTRN at [NBSTRN@nbstrn.org](mailto:NBSTRN@nbstrn.org).

### **NBSTRNs Scope of Work:**

Establish and manage the Newborn Screening Translational Research Network Coordinating Center (NBSTRNCC) and the provision of technical, regulatory, statistical, and administrative support to carry out the tasks specified below, and other tasks as directed by the Project Officer.

1. Establish an organized network of:
  - A. State newborn screening programs
  - B. Clinical centers
2. Develop, implement and refine a national research informatics system for investigators and policy makers that dovetails with established national clinical networks.
3. Establish and administer an efficient and reliable repository of residual dried blood spots that is either virtual or physical. It should be comprised of those stored by State newborn screening programs and other resources.
4. Provide expertise and support related to regulatory requirements associated with informed consent, IRBs, and state and local research policies associated with newborn screening.
5. Facilitate research on the development of new methods and technologies by maintaining close contact with the scientific and biomedical research community.
6. Facilitate research on screened and treated patients to define effectiveness of treatments and long-term outcomes.
7. Provide statistical leadership and clinical trial design expertise for the individualized needs of researchers through the NBSTRN Coordinating Center for procedures.
8. Facilitate the timely dissemination of research findings.
9. Recruit a steering committee comprised of knowledgeable healthcare professionals, public health professionals, ethicists and scientists to make recommendations to Program Officer regarding research proposals to have access to the NBSTRNCC.

Visit our updated website <http://www.nbstrn.org>

## SCID Update

After the acceptance of Severe Combined Immune Deficiency to the Recommended Uniform Screening Panel by the Secretary of the Department of Health and Human Services, the Secretary called for some additional studies. One of these important studies was the NICHD-funded effort, the priorities for this project were to:

- Assess screening technologies for SCID,
- Establish immediate confirmatory tests and procedures for presumed positive results,
- Ensure capacity and resources for tracking positive cases and arrange for appropriate follow-up care and referral in a timely manner, and
- Verify administrative structures necessary for a prospective pilot testing of SCID, including ability to obtain approval for human subject research.

The NBSTRN played several roles during the pilot:

- Administered a subcontract to collect, aggregate, and analyze de-identified screening data generated during the pilot
- Established and hosted monthly conference calls of the pilot investigators and key stakeholders to share information on lessons learned and troubleshoot emerging issues
- Coordinated meetings of SCID experts in NBS, diagnosis, and management
- Standardized diagnosis and evaluation data dictionaries
- Identifying with potential grant applicants to provide preliminary data from pilots to support applications
- Established national network for public health and clinical care

Through this effort over 650,000 newborns have been screened and the investigators working to summarize the results. No missed cases of SCID have come to the attention of the newborn screening programs conducting the pilots.

The SCID pilot has been a successful test of NBSTRN tools, expertise and resources, and we look forward to similar efforts in the future. We invite you to listen in to the monthly calls, held the 4th Friday of every month.

We have our first investigator registered for the VRDBS!

## VRDBS Update

You may have already noticed when visiting to the VRDBS demo site, that there have been some notable changes. The biggest change is the addition of a new section called **Research Support**. This section of the website provides investigators with some new functionality:

- Resource links to grant application and funding sites
- Ability to submit questions to participating state Newborn Screening programs
- Ability to request a VRDBS letter of support

In addition, some design improvements have been implemented to enhance the user experience, such as the **How to Use This Site** page. Check out these and other changes at: <https://nbstrn-demo.5amsolutions.com/tissuelocator-web/browse.action>

## Standing Committee Update

The NBSTRN staff has charged the Standing Committee to refocus the NBSTRN workgroups so they can to produce work products efficiently and effectively, and support the goals of the NBSTRN. The Standing Committee has established 7 key themes for the NBSTRN:

1. Tools/services/expertise
2. Regulation governance "playing nice" within the network-responsible behavior-policy. "rules of engagement"
3. Communication/education
4. Subject engagement and compliance
5. Incentives
6. Facilitating access to data/to network
7. Consenting

Keeping these 7 key themes in mind, the Standing Committee has asked each workgroup to recognize 2 or 3 specific goals and tasks that they will be focusing on in the next two years. The NBSTRN team would like to thank the Standing Committee members for their hard work in the last few months and we look forward to all of the changes. For a full Standing Committee roster, visit our new website!



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