NBSTRN Newsletter

NBSTRN NEWBORN SCREENING TRANSLATIONAL RESEARCH NETWORK

August 2011

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

CONTACT US!

NBSTRN Highlights!

Check out ALL of the NEXT GENERATION MEETING Presentations on http://www.nbstrn.org

Completed Functional Assessment with members from our workgroups. THANK YOU to all that participated!

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

Welcome!!!

Welcome to the first issue of the NBSTRN Newsletter! We are very excited to share with you our updates and highlights every month. This newsletter is designed to keep you informed and identify areas where we can improve our support of researchers.

About the NBSTRN:

In October of 2008, the American College of Medical Genetics (ACMG) was awarded a 5-year contract by the *Eunice Kennedy Shriver* National Institute of Child Health and Human Development (NICHD) to develop the National Coordinating Center for the Newborn Screening Translational Research Network (NBSTRN). The basic goals of our work is to develop a system in which long-term follow-up and outcome data can be collected by providers. This can be used for several purposes including developing the clinical history of the treated disease, some of which can be 1) made available in some form to newborn screening programs as a means of evaluating the success of their screening programs and 2) the basis on which next generation treatments might be evaluated. For conditions that are considered candidates for NBS we will develop a system in which the clinical histories can be defined and in which clinical trials can be pursued.

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

Virtual Repository of Dried Blood Spots (VRDBS)

The NBSTRN is developing a web-enabled repository of de-identified data associated with residual DBS stored in state newborn screening programs. The VRDBS is a tool that facilitates the search for DBS and communication between the investigator and states for retrieval and shipping of DBS specimens for research. A demo version of the VRDBS is now available for review for interested parties and can be accessed through the NBSTRN.ORG website: http://www.nbstrn.org/research-tools. Be sure to check-out the demo site periodically as it is continually updated with improvements, enhancements, and new functionality. Recent enhancements include:

- Homepage look & feel revamped
- Account Registration page updates
- Search DBS page updates
- My Selections page updates
- Proposal Details page updates
- Request Review/Approval page updates
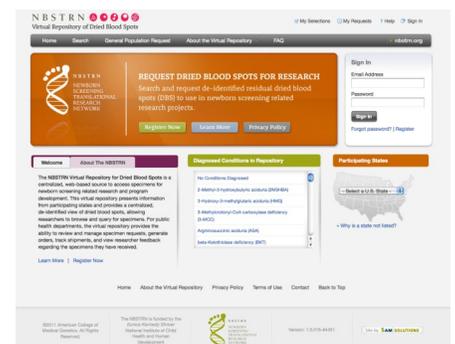
New Research Support in the VRDBS

The NBSTRN team is very excited to develop the new and exciting research support that will be available to investigators through the VRDBS. The Research Support Section will provide an opportunity for investigators that are approved users of the VRDBS to send their questions directly to the states that are participating in the system. State newborn screening programs will be able to answer questions regarding IRBs, DBS specimens, and processes quickly and efficiently. With this new feature, investigators will also be able to request a Letter of Support early in the grant writing process. The researcher will be able to submit his/her request directly to the VRDBS or separately to the NBSTRN by using http://www.nbstrn.org.

The NBSTRN is planning to test this new feature in September by inviting members of the newborn screening community and researchers to complete a functional assessment in which volunteers log into the system and test it's functionality. Through this process, we will be able to fix any bugs and tweak this tool to ensure that it performs as expected.

Stay tuned for more information on this exciting project!

Why LTFU Is So Important

Significant improvements in the health and well-being of newborns affected with a NBS identified disorder have been realized and the majority of these infants now survive into adulthood. Supporting efforts to better understand the factors that contribute to improved health outcomes is an important goal of the NBSTRN. A key component of this effort is the creation of a consensus data set of clinical information that can be collected throughout the life span of affected newborns and aggregated as a resource for researchers, clinicians and public health. The NBSTRN Clinical Centers Workgroup is collaborating on a joint effort to develop this data set with the HRSA supported National Coordinating Center for the Regional Newborn Screening Service Collaboratives (NCC/RC) for Genetics and Long-Term Follow-Up Data Workgroup. This effort lays a strong foundation for a uniform minimum consensus data set to improve understanding of the clinical history of NBS disorders for public health- and research-related activities.

A standard "uniform data set" is being developed that includes demographic and general information that is common to all disorders. Disease specific data sets identify those data elements that are unique to each disorder identified through newborn screening. A web-based tool is also being developed for the capture of this important information.

Workgroup Update

The NBSTRN is comprised of an advisory Standing Committee and four main workgroups made up of national experts, clinicians, consumers, and partner agency representatives. These workgroups meet twice a year in person and hold monthly conference calls. Workgroup members have access to the NBSTRN Workgroup Wiki's where updates, conference call schedules, and work product is posted.

As a result of the NBSTRN Next Generation meeting (June, 2011) each workgroup is prioritizing their goals and tasks for the remainder of the NBSTRN contact. Each month this newsletter will highlight each workgroups and their accomplishments.

Please visit http://www.nbstrn.org to see rosters and learn more about our work.

"Working *together* for a brighter future…"

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