

Where can you see us next?

Public Health & Law Conference
October 10-12, 2012
Atlanta, GA

Annual Meeting of the American Society of Human Genetics (ASHG)
November 6-10, 2012
San Francisco, CA

PRIM&R Advancing Ethical Research Conference
December 4-6, 2012
San Diego, CA

NBSTRN Workgroup Meetings....

NBSTRN Laboratory Workgroup Meeting
September 27-28
Washington, DC
****Note new location****

NBSTRN Bioethics and Legal Issues Workgroup Meeting
October 17-18
Washington, DC

NBSTRN Standing Committee Meeting
December 6-7
Washington, DC

NBSTRN NEWSLETTER



NBSTRN
NEWBORN
SCREENING
TRANSLATIONAL
RESEARCH
NETWORK

August 2012

VRDBS UPDATE

The VRDBS will soon be open to the public with over 2 million DBS specimens available for researchers. This data includes DBS with various data elements including conditions that are a part of the four participating state's newborn screening panel. The NBSTRN is considering including specimens in the VRDBS that are not currently part of the RUSP to further advance the utility of the system. These conditions would be included as "research cohorts": individual specimens identified with conditions not currently in newborn screening for the purpose of developing newborn screening tests. Examples of research cohorts include Fragile X, MPS, and Pompe disease. Further research is required to advance newborn screening for these disorders prior to the conditions being added to the RUSP, and the NBSTRN is working to facilitate this research. The NBSTRN is evaluating the policies, procedures, and consent forms for research cohorts, especially for including specimens from non-participating states. By using the VRDBS investigators will have access to extremely rare and difficult to acquire DBS helping to advance newborn screening and treatments.

Recent Abstracts Accepted for ASHG Meeting

Translational research in newborn screening: implementation of a consensus data set for the long-term follow-up of patients to improve scientific understanding and optimize health outcomes

S. Berry, K. Hassell, A. Brower, Joint Committee of the NBSTRN Clinical Centers Workgroup and NCC/RC Long-Term Follow-Up Workgroup

Translational research in newborn screening: Development of informatics tools to support longitudinal research and accelerate transformations in clinical practice

A. Brower, B. Bowdish, M. Porter, J. Pennington, S. Wrazien, J. Loutrel, P. White, M. Watson

Workgroup Update:

Bioethics & Legal Issues Workgroup

The Bioethics and Legal Issues Workgroup had a very productive workgroup meeting June 5-6th in Chicago. Co-chairs Jeff Brosco and Edward Goldman lead the group on their ongoing work with:

- **IRB Survey:** Written by the workgroup, the survey has been developed in REDCAP and sent out to all of the States. Seventeen states have completed the survey so far. Our goal is to receive results from all 52 (including DC and Puerto Rico) within the next few months. Jeff Botkin will take the lead on data analysis and publication.
- **Incidental Findings Research Topic:** A sub-group was formed to put the finishing touches on a list of "things you should know about incidental findings when working within the NBSTRN". We are hoping to have it complete and on NBSTRN.org by the end of September.
- **VRDBS FAQs:** The workgroup completed their revisions on the FAQs specific to the Virtual Repository. These can be found currently on the demo site (<https://nbstrn-demo.5amsolutions.com/tissuelocator-web/static/faq.jsp>) and will be incorporated into the production site in September.

Due to funding constraints **all** future NBSTRN Workgroup Meetings will be held in the Metro DC area. We apologize in advance for any inconvenience.

Spread the
Word!!!!

R4S Update

LABORATORY QUALITY IMPROVEMENT
OF NEWBORN SCREENING



We have successfully transferred the R4S collaborative project "Laboratory quality improvement of newborn screening" from the servers at Michigan Public Health Institute to the ACMG/ NBSTRN IT environment. It took several weeks to restructure the servers, configure the hardware, and perform comprehensive testing, but thanks to the hard work of our team, we made it happen! Special kudos go out to David McHugh, Gregg Marquardt and Lou Hoffman for putting in those extra hours!

Please update your bookmarks. The new URL is: <http://www.clir-r4s.org/>

Your login credentials haven't changed, but if you've forgotten

