

Where can  
you see us  
next?

Annual SMA  
Conference  
June 13-16, 2013  
Anaheim, CA

**MARK YOUR  
CALENDARS!**

NBSTRN Bioethics  
Workgroup Meeting  
June 6-7, 2013  
Bethesda, MD

NBSTRN Labs  
Workgroup Meeting  
June 27-28, 2013  
Bethesda, MD

# NBSTRN NEWSLETTER

June 2013



N B S T R N  
NEWBORN  
SCREENING  
TRANSLATIONAL  
RESEARCH  
NETWORK

## VRDBS Update

The NBSTRN is excited to announce that the activity in Virtual Repository of Dried Blood Spots (VRDBS) has doubled since our last update. Due to the demos during the NBSTRNetwork meeting in April and recent presentations at the APHL Newborn Screening and Genetic Testing Symposium, we've seen a spike in activity. The VRDBS has twenty two (22) registered investigators, one request for specimens has been completed with another pending, two questions have been submitted and answered by the state programs and one request has been submitted for a letter of support. With some states doing monthly uploads, there is currently data available for over 2.6 million specimens!

Through Google Analytics, the NBSTRN team tracks monthly visitor statistics. For the month of May, we are excited to announce that the VRDBS received 100 new and unique visitors. Since the VRDBS opened in September, the site has had growing numbers, demonstrating the growing success of the VRDBS.

Our VRDBS monthly informational webinars will continue through August as attendance has continued to grow each month. To sign up for one of the next demos, please visit the nbstrn.org site. In addition to the monthly webinars, there are archived sessions available online for those that are unable to make our live demos.

The screenshot shows the NBSTRN website header with the logo and navigation links: Home, About, Research Support, and Search. Below the header is a large orange banner with the text "REQUEST DRIED BLOOD SPOTS FOR RESEARCH" and a description: "Search and request de-identified residual dried blood spots (DBS) to use in newborn screening related research projects." There are three buttons: "Register Now", "Learn More", and "Privacy Policy".

## Spotlight on NBS Researcher

This newborn screening researcher is currently the Deputy Director of the New England Newborn Screening Program and Associate Professor of Pediatrics at the University of Massachusetts Medical School. The principal focus of her work has been the identification and epidemiology of disease that is detectable in neonates through population-based newborn screening. Technical advances from her laboratory have made sophisticated molecular assays available to patients and providers working in domestic centers and international centers that would otherwise be unable to access such technology.

Read more and find out who the Researcher of the Month by visiting:

<https://www.nbstrn.org/about/spotlight/Comeau>

## Pompe Disease Added to RUSP

The US Secretary of Health and Human Services' Discretionary Advisory Committee for Heritable Disorders in Newborns and Children (DACHDNC) voted in May to add Pompe Disease to the recommended uniform newborn screening panel (RUSP). The recommendation next goes to the Secretary for her action.

Currently, Missouri is the only state screening for Pompe. In a pilot project that began on January 15, 2013, they have identified 1 case of classic infantile Pompe Disease to date. Information presented at the DACHDNC predicted a probable incidence of about 1:28,000 (including late onset disease) in the US based on limited studies to date. While a recommendation by the DACHDNC does not have legal standing since a national newborn screening program in the US does not exist, most state programs consider the recommendations seriously and implementation is usually only a matter of time.

## NBSTRN Staff Transition

It is with mixed emotions that we announce that Amy Hoffman, the Project Manager for NBSTRN has accepted a position with the Seattle Children's Hospital as the Senior Program Manager for the Cystic Fibrosis Therapeutics Development Network (TDN). She will be leaving the NBSTRN effective June 10<sup>th</sup> and will begin transitioning out with July 19<sup>th</sup> being Amy's last day. Irina Smotrich will take over the Project Manager duties until the contract is renewed. Please contact her ([ismotrich@acmg.net](mailto:ismotrich@acmg.net)) after June 10<sup>th</sup> for any NBSTRN-related issues.

The NBSTRN is excited for Amy and her family and thank her for all of her hard work for the past four years. The NBSTRN team will miss her!

If you have topics for inclusion in future newsletters, please let us know by emailing [ismotrich@acmg.net](mailto:ismotrich@acmg.net)



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