

# NBSTRN NEWSLETTER



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NEWBORN  
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## MARK YOUR CALENDARS!

### NBSTRN Monthly Webinars!

A Demonstration of  
the VRDBS

July 18th, 2013,  
2:00pm EST.

### Have you seen the *Providing New- born Screening Specimens for Research Webinar Series?*

Visit [NBSTRN.org](http://NBSTRN.org) to  
download!

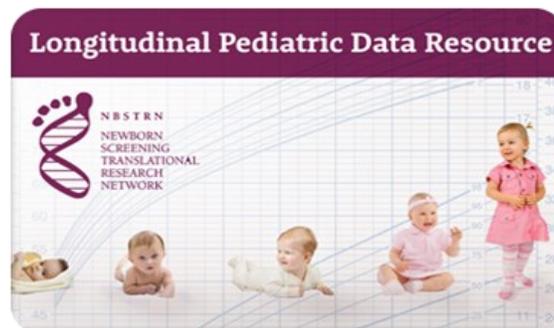
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July 2013

## Use of the LPDR to Support Translational Research in Newborn Screening

The Longitudinal Pediatric Disease Resource (LPDR) is a combined system of electronic data capture, centralized data management, unified data representation, and case reporting within a FISMA-compliant architecture. Currently the LPDR includes health information for 46 metabolic conditions, five lysosomal storage disorders and one neuromuscular disorder. This web-based system allows clinicians to access electronic disease specific electronic case report forms (CRFs) that incorporate a uniform dataset as well as disease-specific datasets that were developed through a national consensus process. Data can be collected as part of a standard clinic visit and entered into a centralized or institutionally-enabled REDCap instance for aggregation, management and analysis. Three research projects are currently using the LPDR. One research team is actively using the LPDR to better understand the natural histories of rare metabolic disorders that are part of routine newborn screening and to verify the effectiveness of early identification, intervention and treatment. Two research teams are using the LPDR to study conditions that have been proposed for newborn screening. These studies will work to determine the clinical and diagnostic accuracy of screening assays, investigate how to correctly predict phenotypes in asymptomatic newborns, and develop algorithms to assist with clinical decision-making about if and when to initiate therapy. In addition, these studies will explore novel ethical, legal, and social issues associated with testing infants for potentially later-onset disorders. The LPDR is available to the newborn screening research community and it is expected to accelerate the development of new technologies and treatments, methodological, outcomes, and public health research of newborn screening.



## Spotlight on NBS Researcher

This month's spotlight researcher is recognized for his work as the director of a State Department of Public Health's Genetic Disease Screening Program. He received his PhD in Biological Anthropology and Genetics from the University of California, Davis and specializes in human genetics and genetic disease epidemiology. He is well known to the newborn screening research community for his knowledge and experience in public health, protein electrophoresis and chromatography.

Our mystery researcher of the month oversees research on morbidity and mortality for all infants, which can be found through screening programs based on treatment compliance, genetic haplotypes, demography, and multi-disciplinary services at specialty centers. In addition, he has brought new insight into the investigation of the possible correlation between perchlorate spill and hypothyroidism, MCADD and SIDS, as well as SIDS and sickle cell traits. He has been a motivating force behind the pilot and implementation of SCID in his State. He currently serves on the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children.

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

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

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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## Bioethics and Legal Issues In-Person Meeting Update

We would like to take this opportunity to say "thank you" to our Bioethics and Legal Issues Workgroup for their hard work at the face-to-face meeting on June 6<sup>th</sup> and 7<sup>th</sup>.

The Bioethics & Legal Workgroup had a productive meeting focusing on several key points. The meeting began with an update on the Longitudinal Pediatric Data Resource (LPDR) which led to an informative discussion on data sharing and access. The group made recommendations regarding the Levels of Access for users entering the Longitudinal Pediatric Data Resource (LPDR) and gave excellent feedback on the Terms and Conditions specific to the LPDR tool. In addition to the wonderful work with the LPDR, the workgroup reviewed and edited the summary for the "Providing Newborn Screening Specimens for Research: Webinar Series", which will soon be available on the NBSTRN website.

We concluded our meeting with a reflection of the last five years and the goals which the Bioethics Workgroup has met or exceeded, and those goals which it still wishes to accomplish. With new issues constantly coming to the forefront in research, NBSTRN is glad to have the opportunity to work with the individuals on the Bioethics and Legal Issues Workgroup.