

Where can  
you see us  
next?

Gatlinburg Conference  
March 6-8, 2013  
San Antonio, TX

ACMG Annual Meeting  
March 19-23, 2013  
Phoenix, AZ

2013 Joint Meeting of  
the Newborn Screen-  
ing and Genetic  
Testing Symposium  
and International  
Society for Neonatal  
Screening  
May 5-6, 2013  
Atlanta, GA

### MARK YOUR CALENDARS!

NBSTRN Clinical  
Centers Executive  
Committee Meeting  
April 4-5, 2013  
Washington, DC

NBSTRN Network  
Meeting  
April 8-9, 2013  
Washington, DC

NBSTRN Standing  
Committee Meeting  
April 7th & 9th, 2013  
Washington, DC

# NBSTRN NEWSLETTER



N B S T R N  
NEWBORN  
SCREENING  
TRANSLATIONAL  
RESEARCH  
NETWORK

March 2013

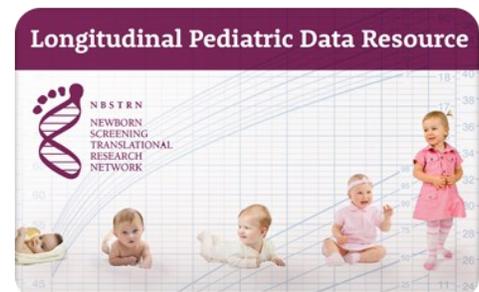
## Longitudinal Pediatric Data Resource (LPDR)

The Newborn Screening Translational Research Network (NBSTRN) has developed the **Longitudinal Pediatric Data Resource (LPDR)**, a combined system of standardized data elements, electronic data capture, centralized data management, unified data representation, and case reporting within a FISMA-compliant architecture. The LPDR is available for researchers, clinicians, and public health teams to use for secure data collection, sharing, management, and analysis across newborn screening conditions while establishing a valuable resource for the community.

### FEATURES of the LPDR:

- Data Sets – developed by a national consensus process, these common data elements, publicly available, to incorporate into public health, clinical, basic and translational efforts across different conditions (currently 48 different disorders)
- Informatics System – an informatics system to enable enhanced data collection, data management, case reporting, and data analysis that utilizes REDCap (Research Electronic Data Capture) electronic data capture tools for prospective, longitudinal research
- Standardization – a data almanac of definitions, annotations and standards for each data element where available
- Pilots - the LPDR is currently being used by several grantees investigating the natural history of newborn screening conditions and piloting newborn screening for new conditions including spinal muscular atrophy and 5 lysosomal storage disorders

Visit [www.NBSTRN.org](http://www.NBSTRN.org) or contact  
us at [nbstrn@nbstrn.org](mailto:nbstrn@nbstrn.org) for more  
Information!



## Spotlight on NBS Researcher

This researcher of the month is a Senior Physician in Medicine at Children's Hospital Boston and a Professor of Pediatrics at Harvard Medical School. He was formerly the Director of the Metabolic Program at Children's Hospital Boston and currently serves as an attending physician within the program.

A strong advocate of the expansion of newborn screening, he was instrumental in Massachusetts becoming the first state to include 20 additional metabolic disorders in its newborn screening program.

Not sure who we are talking about yet? Go to: <https://www.nbstrn.org/about/spotlight/Levy> to read more!

## NCC Community Conservation

### Emerging from Darkness to Shed Light on Genetics and Cultural Taboos in Native American Populations: A Community Conversation on March 19<sup>th</sup> at the ACMG Annual Meeting in Phoenix, AZ

The tension between the potential knowledge that can be gained through genetics research and medicine, and cultural beliefs within the Native American Populations of the United States, was explored at the inaugural Community Conversation held at the 2009 ACMG Annual Meeting ("Genetics and Genetic Research: Native American Perspectives".) With the release of the documentary, "Sun Kissed," (<http://sunkissedthefilm.com/>) these tensions are further highlighted as one family, the Nez's, works to understand why their children were born with Xeroderma Pigmentosum (XP) and why the prevalence within the Navajo community is 1 in 30,000 versus 1 in one million in the general population. "Sun Kissed" follows the Nez's as they negotiate between the cross-cultural norms of their community and their information and support needs as parents of children with an incurable genetic condition.

At the ACMG 2013 Annual Meeting in Phoenix, AZ, this special satellite session will provide ACMG meeting attendees an opportunity to view the documentary "Sun Kissed" and participate in a panel discussion with the Nez's and the filmmakers. ACMG conference participants are invited, as are families, health care providers, public health professionals, the media and **YOU** to join the conversation. Free drinks and popcorn will be served!



**March 19<sup>th</sup> at 7:00pm**  
**ACMG Annual Meeting in Phoenix, AZ**

This session is sponsored by the National Coordinating Center for the Genetic and Newborn Screening Services Collaboratives (NCC) and the Mountain States Genetics Services Collaborative (MSGRC). The NCC is funded by U22MC04100, awarded as a cooperative agreement between the Health Resources and Services Administration/Maternal and Child Health Bureau/Genetic Services Branch (HRSA/MCHB/GSB) and ACMG. The MSGRC is funded by H46MC24095 as a grant from the HRSA/MCHB/GSB to the Texas Health Institute.

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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