



NBSTRN NEWSLETTER

Upcoming Meetings for NBSTRN:

APHL Newborn
Screening and Genetic
Testing Symposium
Feb 29-Mar 3,
St. Louis, MO

LSD World
Symposium
Feb 29-Mar 4
San Diego, CA

ACMG Annual Clinical
Genetics Meeting
Mar 8-12, Tampa, FL



Consider taking the... ***NBSTRN Researcher Needs Assessment***

Survey Overview: The Newborn Screening Translational Research Network ([NBSTRN](#)) would like ensure that the needs of newborn screening researchers are being met and to see to what improvements the NBSTRN may contribute to the newborn screening research community. By participating in this brief needs assessment, you will provide valuable information to NBSTRN and be informed about numerous tools and resources that we offer.

NBSTRN's History: The NBSTRN is a contract funded by the *Eunice Kennedy Shriver* National Institutes of Health (contract # HHSN275201300011C) awarded to the American College of Medical Genetics and Genomics (ACMG). Throughout its contract, the NBSTRN has worked to encourage, assist and expand newborn screening research programs. The NBSTRN supports newborn screening researchers, including the public health and epidemiological researchers who use dried blood spots that some states retain. The NBSTRN's mission is to improve the health outcomes of newborns with genetic or congenital disorders by means of an infrastructure that allows investigators access to robust resources for newborn screening research.

Survey responses will be aggregated and not individually identified. Your responses will be kept anonymous. The survey should take ten to fifteen minutes to complete.

Please [click here](#) to access the survey.

If you have any questions, please email Rachel Fleming (rfleming@acmg.net).

Thank you for taking the time to provide your thoughts and feedback,

The NBSTRN team





Spotlight on a Newborn Screening Researcher

The first spotlight researcher of 2016 has been the Director of the Colorado Sickle Cell Treatment and Research Center at the University of Colorado since 2003. In addition, she was serving as its Associate Director since 1992. She is also a Professor of Medicine at the University Of Colorado School Of Medicine.

This mystery spotlight researcher earned her Medical Degree from the University of Minnesota Medical School in Minneapolis, MN. Her clinical research interests include Coagulation disorders and anticoagulation management, hemoglobinopathies including sickle cell disease and hematology support of the Bloodless Surgery Program.

Currently she helps provide inpatient and outpatient care to adults with sickle cell disease at the University of Colorado Hospital and is a site principal investigator for multi-center clinical trials for adults with sickle cell disease. She has led several regional projects through the Mountain States Genetics Regional Collaborative Center (MSGRC), and serves on the Newborn Screening Advisory Committee for Colorado State Department of Health.

To find out more on how else she has contributed to newborn screening, visit: <https://www.nbstrn.org/about/spotlight-researchers>

Natural History of Disorders funding

The Newborn Screening Saves Lives Reauthorization Act of 2014 authorizes the Hunter Kelly Newborn Screening Research Program within NICHD to carry out, coordinate, and expand research in newborn screening. As a part of this program, NICHD recently announced a new funding opportunity to develop a comprehensive description of the natural history of disorders that could potentially benefit from newborn screening or currently screened conditions that could benefit from expanded knowledge. This effort will enable assessment of the effectiveness of treatments and interventions used across the lifespan for infants and children identified via newborn screening. It will also facilitate understanding of the genetic and clinical heterogeneity of conditions that are candidates for routine newborn screening, and inform the approach and timing of age-appropriate clinical care.

For more information on this new opportunity please visit <http://grants.nih.gov/grants/guide/pa-files/PAR-16-061.html>.

If you have topics for inclusion in future newsletters, please let us know by emailing

rfleming@acmg.net



NBSTRN

Newborn Screening
Translational Research
Network