

## NBSTRN NEWSLETTER

### Upcoming Meetings for NBSTRN:

**APHL Newborn  
Screening and Genetic  
Testing Symposium**

**Feb 29-Mar 4,  
Washington DC**

**LSD World  
Symposium**

**Feb 29-Mar 4  
San Diego, CA**

**ACMG Annual Clinical  
Genetics Meeting**

**Mar 8-12, Tampa, FL**



## Comment period extended for the NPRM on Revisions to the Common Rule

A notice of proposed rulemaking (NPRM) was released by the U.S. Department of Health and Human Services (HHS) in early September 2015 to announce proposed revisions to the regulations protecting human subjects in research.

**The period to comment** on the NPRM has been extended by 30 days with a new deadline of January 6th, 2015 at 11:59pm EST. If you wish to comment, you can follow the instructions provided on the OHRP webpage entitled "[NPRM for Revisions to the Common Rule](#)" that is also accessible from the [OHRP home page](#). We have also provided the instructions below:

It is noted that you can submit in three ways (no duplicates, please):

**1. Electronically (online).** You may submit electronic comments on specific issues in this regulation. (Attachments should be in Microsoft Word, WordPerfect, or Excel; however, we prefer Microsoft Word.)

*In another tab or window, access [www.regulations.gov](http://www.regulations.gov):* The docket number is HHS-OPHS-2015-0008, which can be typed into the [regulations.gov](#) box that is labeled "Search". The first answer should say "Featured Result." Click on "Comment Now" button to the right of the notice title.

*Or use this direct link to the comment box:* <http://www.regulations.gov/#!submitComment;D=HHS-OPHS-2015-0008-0001>

**2. By mail.** You may mail written comments (one original and two copies) to the following address ONLY: Office for Human Research Protections (OHRP), 1101 Wootton Parkway, Suite 200, Rockville, MD 20852. Please allow sufficient time for mailed comments to be received before the close of the comment period.

**3. By hand or courier.** If you prefer, you may deliver (by hand or courier) your written comments (one original and two copies) before the close of the comment period to the Office for Human Research Protections (OHRP), 1101 Wootton Parkway, Suite 200, Rockville, MD 20852.



## Spotlight on a Newborn Screening Researcher

December's spotlight researcher was a Professor and Geneticist of Pediatrics at The University of Texas Health Science Center at San Antonio. She was also a Professor and Senior Associate Dean for Education, Office of Educational Development and Research at the University of Colorado, School of Medicine until her recent retirement of last year. Additionally, she served as the Project Director of the Mountain States Genetic Services Regional Collaboratives (MSGRC).

She began her education at Wayne State University in Detroit, Michigan earning both her BS and MD degrees, and later went on to receive her PhD from Northwestern University. Our mystery researcher focuses her research on medical education, inborn errors of metabolism, and newborn screening. She has been the principle investigator for over 25 federally and privately funded projects. She has helped serve as the senior advisor and member of the National Newborn Screening and Genetics Resource Center Steering Committee. She was also a member of the Centers of Disease Control and Prevention's Evaluation of Genomic Applications in Practice and Prevention Workgroup (EGAPP) and the American Academy of Pediatrics Working Group on policy for Newborn Screening and Medical Home.

Furthermore, she has been an author for many peer reviewed publications that have helped with the enhancement and understanding of genetic services, newborn screening, and more. To find out more on how else she has contributed to newborn screening, visit: <https://www.nbstrn.org/about/spotlight-researchers>

## Newborn Screening for MPS-1 Pilot Study

This past May, the *Eunice Kennedy Shriver* National Institute of Child Health and Human Development at the National Institutes of Health issued a Request for Proposal for a Newborn Screening Pilot Studies that was awarded to multiple organizations in late September and early October. The award notices were distributed to three high throughput laboratories for an MPS-1 Pilot Study. The three organizations were chosen by their ability to support the development, implementation and proof of concept of newborn screening for MPS-1.

The awardees; Emory University, University of Massachusetts, Worcester, and Research Triangle Institute, North Carolina have since begun to collaborate with the NBSTRN on conference calls and plan to leverage the resources available through the NBSTRN, including access to the NBSTRN's Laboratory Performance Program which was developed to collect, aggregate and analyze de-identified screening data. If you would like to find out more about the NBSTRN's Laboratory Performance Program, the R4S, you can find more information on the [NBSTRN.org](https://www.nbstrn.org) or the [R4S landing page](#).



**NBSTRN**

Newborn Screening  
Translational Research  
Network

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