

NBSTRN NEWSLETTER



N B S T R N
NEWBORN
SCREENING
TRANSLATIONAL
RESEARCH
NETWORK

Where can you see us next?

AMCHP Meeting
February 9-12, 2013
Baltimore, MD

WORLD Symposium
February 12-15, 2013
Orlando, FL

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 Network
Meeting
April 8-9, 2013
Washington, DC

February 2013

Upcoming NBSTRN Posters and Presentations

The NBSTRN Coordinating Center team will be presenting posters and oral presentations at several upcoming meetings.

Meeting	Title
AMIA Joint Summits on Translational Science March 18-22	The Development of the Longitudinal Pediatric Disease Resource to Support Translational Research in Newborn Screening
ACMG Annual Meeting March 19-23	Providing Residual Newborn Screening Specimens for Research: Legal and Ethical Implications to Consider
ACMG Annual Meeting March 19-23	Development of the Longitudinal Pediatric Disease Resource (LPDR)
ACMG Annual Meeting March 19-23	Use of the Longitudinal Pediatric Disease Resource (LPDR) by the Inborn Errors of Metabolism Collaborative (IBEMC)
ACMG Annual Meeting March 19-23	Development and Implementation of a Tool for the Clinical Validation of Severe Combined Immune Deficiency (SCID) Newborn Screening
APHL NBS and Genetic Testing Symposium May 5-10	Providing Residual Newborn Screening Specimens for Research: Legal and Ethical Implications to Consider
APHL NBS and Genetic Testing Symposium May 5-10	Translational Research in Newborn Screening: Development of a Virtual Repository of Residual Dried Blood Spots
APHL NBS and Genetic Testing Symposium May 5-10	Translational Research in Newborn Screening: Efforts to Facilitate Implementation of Newborn Screening for Severe Combined Immune Deficiency (SCID)
APHL NBS and Genetic Testing Symposium May 5-10	Development of Informatics Tools to Support the Long-Term Follow-Up of Individuals with Conditions Included in Newborn Screening: A Pilot Focused on Inborn Errors of Metabolism
IDF National Conference June 27-29	SCID Newborn Screening

Spotlight on NBS Researcher

He is the author of over 150 papers with genetic-metabolic diseases in childhood. He served as Senior Advisor to the Director, Eunice Kennedy Shriver National Institutes of Child Health and Human Development at the NIH in Bethesda, MD (2004-2011). During this time he oversaw some of the NICHD research initiative in newborn screening, which is by far the most common use of genetic testing today. He was also the Founding Chair of the Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children.

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

Staff Update

The NBSTRN Coordinating Center is excited to announce that Rachel Fleming has joined our team as the new administrative assistant. Rachel Fleming graduated in 2011 from St. John's College in Annapolis, Maryland where she pursued a degree in liberal arts. With three years of laboratory science classes at St. John's College, Rachel became increasingly interested in science education and pursued a career as a middle school science teacher. She enjoyed her time teaching; however, in order to learn more about public health and the role of ethics in the medical field she traded in her teaching career for a position at ACMG as an administrative assistant. She has recently been brought on to the NBSTRN team and hopes to keep learning more about these areas of interest. Welcome Rachel!

Fun fact: Her favorite scientist is Alexander Fleming, duh!

VRDBS Update

The Virtual Repository of Dried Blood Spots has been operating without any setbacks since September, 2012 and we are excited to announce that:

- New states will move immediately to productions without a pilot
- Over 2 and a half million DBS
- 15 registered investigators (2 more pending)
- First request made (review pending)

You can find out more about the VRDBS by attending one of our monthly public demonstrations. Go to <https://www.nbstrn.org/research-tools/virtual-repository-of-dried-blood-spots/demo-dates> to register.

Future demo dates include:

2/21/13 3/21/13 4/18/13 5/16/13 6/20/13

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

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