Newborn Screening Translational Research Network

October 2014

NBSTRN NEWSLETTER

Upcoming Meetings for NBSTRN:

2014 Public Health Law Conference

Oct 16-17, Atlanta, GA

2014 ASHG Annual Meeting

Oct 18-22, San Diego, CA

2014 Newborn Screening and Genetic Testing Symposium

Oct 27-30, Anaheim,

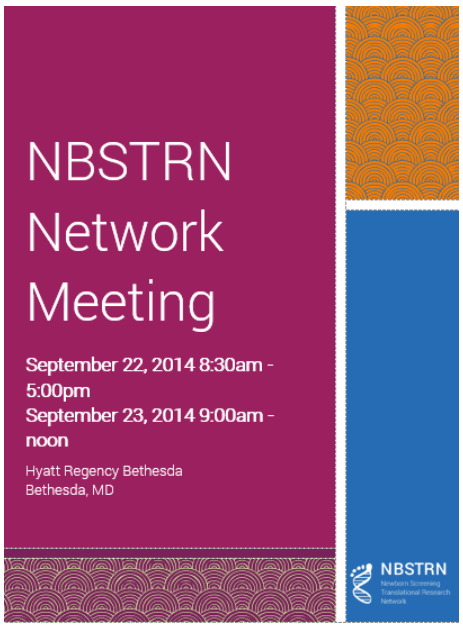
NBSTRN Network Meeting

We'd like to extend a warm "thank you" to all who participated in making this meeting a great success! We had a wonderful turn out for our Network Meeting on September 21st and 22nd. Of the 91 people registered, 83 were able to join us.

We learned about the various tools and resources available through the Newborn Screening Translational Research Network (NBSTRN), heard from grantees working with the NBSTRN tools and had the opportunity to learn about ongoing and future pilots in newborn screening. In addition, we heard presentations on the ethical and legal implications associated with doing newborn screening as well as the future of next generation sequencing. To conclude the meeting, we had the opportunity to hear from federal partners who told us about many different research opportunities.

Overall, it was a very productive day and a half and will help us to prepare for the next phase of the NBSTRN. We look forward to continuing our efforts in perfecting our tools and supporting newborn screening researchers.

In order to improve our efforts, we've developed a brief feedback survey. Please visit http://surveys.acmg.net/redcap/surveys/?s=m9DJGf to provide us your feedback on the Network Meeting.



NBSTRN is funded by a contract from the *Eunice Kennedy Shriver* National Institute of Child Health and Human Development, National Institutes of Health to the American College of Medical Genetics and Genomics (#HHSN275201300011C).

Spotlight on a Newborn Screening Researcher

Our spotlight researcher of the month is currently a clinical, biochemical and molecular geneticist in the division of medical genetics at Emory University, School of Medicine. He specializes in the diagnosis and treatment of metabolic disorders and genetic disorders of the skeleton, particularly dwarfisms and limb deficiency disorders. He is board certified by the American Board of Medical Genetics in clinical genetics, clinical biochemical genetics, and clinical molecular genetics.

This researcher has an extensive research background in clinical, biochemical and molecular genetics. His current research focuses on clinical trials of therapies for genetic disorders, particularly lysosomal storage disorders. He also researches the genetic basis and pathology of genetic disorders of the skeleton, particularly skeletal dysplasias and limb deficiency disorders. He has co-authored or contributed to over one hundred articles in his time as a clinical geneticist.

Read more and find out who the Researcher of the Month is by visiting: https://www.nbstrn.org/about/spotlight-researchers

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

rfleming@acmg.net

NBSTRN Steering Committee and LSD Workgroup meeting updates

In conjunction with the NBSTRN Network Meeting, the NBSTRN Steering Committee met on September 23rd for half a day to discuss the NBSTRN Year 1 goals and accomplishments. The committee reviewed its recommendations for the NBSTRN from the Spring 2014 meeting and made new recommendations and set goals for year 2 of the NBSTRN.

The NBSTRN Lysosomal Storage Disorder (LSD) Workgroup met for a one-day meeting on September 24th. The goal of the meeting was to review and complete the disease specific common data elements (CDE's) for Pompe, Gaucher, Fabre, Niemann-Pick AB, and Pompe disease. The workgroup also heard about various ongoing newborn screening efforts for LSD's. This workgroup will continue its efforts with regular conference calls .

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