Newborn Screening Translational Research Network

December 2014

NBSTRN NEWSLETTER

Upcoming Meetings for NBSTRN:

AMCHP 2015
Jan 24-27, Washington DC

LSD World Symposium
Feb 9-13, Orlando, FL

Clinical Integration Group Face-to-Face meeting
Feb 2-3, Bethesda, MD

NCC Celebrates 10 Years at ACMG

The National Coordinating Center for the seven HRSA's Regional Genetic Services Collaboratives, collectively known as the NCC/RC system, has resided at the American College of Medical Genetics and Genomics (ACMG) since the system's inception in 2004. Currently, the NCC focuses on evaluating the work of the seven RCs, pursuing unique projects of national interest, and in building bridges between genetics and local communities.

In 2013, the NCC's national evaluation program conducted national surveys in collaboration with the National Genetics Education and Consumer Network (NGECN, part of NCC), the seven RCs, and the ACMG membership. These national surveys were the first of their kind to ask at a national level about medical home, care coordination, and transition from genetics providers and consumer perspectives. These data formed the baseline to current NCC/RC activities and two publications are forthcoming. These surveys will be fielded again in winter 2015.

NCC's national projects focus on the genetic component of electronic medical records (EMR), ACT Sheet development and maintenance (in partnership with ACMG), and in developing public health data collection for long-term follow-up (LTFU). The EMR project is working with ACMG's Economics of Genetic Services Committee and the Institute of Medicine's Genetics Roundtable to ensure that the work of NCC aligns with that of ACMG and IOM. The EMR project will hold a meeting in the spring 2015. The ACT Sheet workgroup is currently addressing secondary findings and has a draft set of 13 sheets in development. Finally, the LTFU workgroup has a set of public health elements ready for pilot in state follow-up programs.

The NCC welcomes those who are interested to participate and contribute to the NCC/RC system. For more information, please go to www.nccrcg.org or contact Alisha Keehn, akeehn@acmg.net.

Alisha Keehn, MPA, NCC Project Manager

NCC
National Coordinating Center
for the Regional Genetic Service Collaboratives

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Spotlight on a Newborn Screening Researcher

Our spotlight researcher is currently the director of the Phenotyping Core of the Program in Genomics at Boston Children's Hospital. She oversees the interactions between the Gene Discover Core (GDC) participants. Her research focuses on the genetics of bone disorders. She has performed research in mutations of the PHEX gene in hypophosphatemic rickets. She is particularly interested in the contribution of environmental factors to the low bone mass seen in individuals with neuromuscular diseases, such as muscular dystrophy.

As director of the Phenotype Core of the Program in Genomics, she has worked with researchers to develop clinical genetic research projects in diabetes, autism, congenital heart disease, and atopic dermatitis, and has her own project in the genetic contributions to congenital hip dysplasia.

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

Newborn Screening Pompe Pilot Workgroup Conference Calls

This 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 Pompe Pilot Study that would likely be awarded to multiple organizations starting September 26th, 2014 for an 18-month period of performance. In September, the award notices were distributed to three organizations that would support the development, implementation and proof of concept in newborn screening for Pompe Disease .

The awardees; Emory University, the Wisconsin State Laboratory of Hygiene, and the Health Research Inc. in New York have since begun to collaborate with the NBSTRN on bi-monthly 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 or the R4S landing page.

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