Newborn Screening Translational Research Network

May 2014

NBSTRN NEWSLETTER

Upcoming Meetings for NBSTRN:

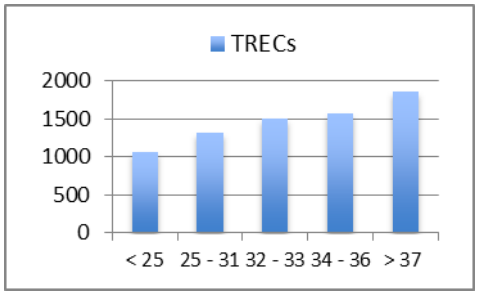
Bioethics and Legal Issues Workgroup Face-to-Face Meeting
May 14th-15th
Bethesda, MD

Steering Committee Face-to-Face Meeting
May 15th-16th
Bethesda, MD

New York State Report on SCID Screening

In 2010 New York State, under the direction of Dr. Michele Caggana, led a national pilot of SCID newborn screening that screened over 654K babies and established several key resources including high-volume newborn screening protocols, laboratory and clinical follow-up algorithms, and protocols for treatment.[1] Dr. Caggana's team now reports on the first two years of statewide screening in New York State in the April edition of the Journal of Clinical Immunology.[2] Beth Vogel, the lead author, reports several key findings based short and long-term follow-up data on the 485,912 infants screened including:

- 10 infants with SCID were identified, 90% received a transplant or enzyme replacement therapy and are doing well
- SCID incidence of approximately 1 in 48,500 births is consistent with other screening states but higher than reports before newborn screening (1 in 100,000)
- Incidence of clinically significant laboratory abnormalities of approximately 1 in 5,000
- Statistically significant findings are summarized in the table below:

Finding	Details	p value
More males than females referred	1.72	p<0.001
Increased frequency of Black infants referred for low TREC values		p<0.001
Number of TRECs varies by gestational age		p<0.001

The report recommends continued national collaboration to understand the long-term impact of SCID newborn screening and encourages the use of the NBSTRN and other partners to assess laboratory practices, diagnosis, short- and long-term follow-up.
Amy Brower, Ph.D.

Sources:
1. https://www.nbstrn.org/facilitated-researchs/severe-combined-immunodeficiencies
2. Vogel, BH, et al. Newborn Screening for SCID in New York State: Experience from the First Two Years; 2014; Apr;34

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Spotlight on a Newborn Screening Researcher

Our spotlight researcher of the month is currently the Director of the Center for Pediatric Genomic Medicine at Children's Mercy Hospital in Kansas City, Missouri and Professor in the Departments of Pediatrics and Obstetrics & Gynecology at the University of Missouri, Kansas City. As the Director of the Center for Pediatric Genomic Medicine at Children's Mercy, he and his team have developed a genome sequencing test for newborns and children that scans for 600 severe genetic diseases and provides quick results within four to six weeks.

This researcher has an extensive background in the field of genetic research. He received his educational degrees primarily from Queens University in Belfast, Northern Ireland. At Queens, he completed a Bachelor of Science degree, Bachelor of Medicine degree, Bachelor of Surgery degree and a Bachelor of the Art of Obstetrics degrees. He went on to complete a postdoctoral degree in genetics at Duke University and became an Assistant Professor in the department of medicine at the Center for Mammalian Genetics. In 2010, he received his doctorate of science degree and specializing in molecular genetics. From then on he has focused his work on genome analysis for inherited diseases.

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

NBSTRN Booth at the ACMG Meeting

This year marked the first time that the NBSTRN booth exhibited in the ACMG Annual Meeting's Exhibit Hall. The NBSTRN Booth provides a great opportunity for researchers to stop by and meet with knowledgeable staff members and learn more about the NBSTRN tools and resources that are available to researchers and grantees.

At the ACMG Annual Meeting, we were able to showcase a new addition that has been added to the NBSTRN Booth. Meeting goers who are curious about how to register for NBSTRN tools and features now have the ability to explore our website and research tools on our new exhibit booth Tablet. At all future exhibit booths to come, we will be displaying different features of NBSTRN tools such as the Longitudinal Pediatric Database's (LPDR) electronic Case Report Forms (eCRF's), or the very easy multiple tool registration process. Look for the NBSTRN Booth at future national meetings and we will be happy to inform you about our tools.

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

rfleming@acmg.net

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