Newborn Screening Translational Research Network

February 2014

NBSTRN NEWSLETTER

The VRDBS has more investigators every month! If you are interesting to find out how to become an investigator, visit the VRDBS homepage

Lysosomal Storage Diseases

Lysosomal storage diseases (LSDs) are a group of distinct genetic disorders characterized by a specific defect in a lysosomal enzyme that causes the accumulation of glycolipids. Most LSDs are progressive and often result in severe morbidity and early death with an overall incidence of 1 in 7000 to 1 in 8000 live births.[1] Newborn screening for several lysosomal storage disorders has been proposed in several states and the Discretionary Advisory Committee on Heritable Disorders in Newborns and Children (DACHDNC) has received nominations for routine screening for Krabbe, Fabry, Niemann Pick and Pompe. The Pompe nomination was referred for an evidence review and subsequently the DACHDNC recommended addition of Pompe to the Recommended Uniform Screening Panel (RUSP) in May 2013.[2] Pompe-causing mutations are present in about one in 40,000 newborns and the infantile-onset form of the disease affects about one in 133,000 infants. The late-onset form of the disease affects from one in 26,466 to one in 57,000 individuals. Pilots of newborn screening for Pompe are ongoing in several states including Missouri and New York. NBSTRN is supporting the pilots by providing use of the VRDBS for biospecimen sharing, the R4S for test development, and the LPDR for collection of prospective longitudinal clinical information. Disease specific common data elements have been drafted by the NBSTRN LSD Workgroup and are available for researchers, clinicians and newborn screening programs to incorporate into their efforts. Please contact us if we assist you in your efforts to better understand the LSDs.

1. Meikle PJ, Hopwood JJ, Clague AE, Carey WF. Prevalence of lysosomal storage disorders. JAMA. 1999;281:249–54

2.http://www.hrsa.gov/advisorycommittees/mchbadvisory/heritabledisorders/nominatecondition/reviews/pompereport2013.pdf

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).

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

ibutler@acmg.net

Spotlight on a Newborn Screening Researcher

Our monthly researcher brings his background in microbiology and immunology to the world of newborn screening. He received his master's and doctorate from the State University of New York and before that he received his bachelor's degree from the University of Vermont. Afterwards, he completed his postdoctoral work at Harvard University and has since become the Director of the UMMS New England Newborn Screening Program (NENSP) at the Center for Health Policy and Research.

Since he joined UMMS, this researcher has taken a particular interest in the congenital toxoplasmosis screening. In 1999, his team at NENSP became one of the first groups in the country to apply tandem mass spectrometry to state newborn screening tests. This researcher also helped coordinate efforts to add succinylacetone screening in 2008 as well as screening for severe combined immunodeficiency (SCID) in 2009.

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

ACMG Welcomes New Medical Director

ACMG will welcome David B. Flannery, MD, FACMG, FAAP as its new Medical Director next month. Dr. Flannery is a Founding Fellow of the College and has served on the ACMG's Economics of Genetics Services Committee (member and co-chair), Professional Practice and Guidelines Committee (member), and CPT and Reimbursement Committee (member). He was on the Editorial Panel for the ACMG's *Genetics Billing and Reimbursement Manual* and he is currently the ACMG representative to the American Medical Association's CPT Advisory Committee. He has also served as a member of the Southeast Regional Newborn Screening & Genetics Collaboratives (SERC). SERC is one of the seven HRSA Genetics Collaboratives that is under the National Coordinating Center, housed at ACMG through its cooperative agreement with MCHB/HRSA.

Please watch for a formal, comprehensive announcement in February about Dr. Flannery who is succeeding Dr. Barry Thompson, who recently retired after five years of dedicated work to ACMG.

NBSTRN
Newborn Screening Translational Research Network

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).