Newborn Screening Translational Research Network

November 2015

NBSTRN NEWSLETTER

Mark your calendars!

APHL NBS and Genetic Testing Symposium

Feb 29– Mar 3, St. Louis, MO

ACMG Annual Meeting

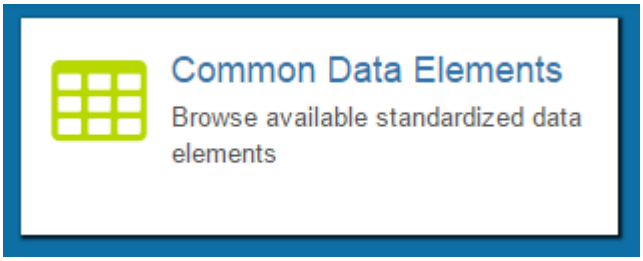
Mar 8-12, Tampa, FL

Lysosomal Storage Disorders (LSD) Workgroup Update

Researchers across the country are conducting ground breaking studies using NBSTRN resources to further understanding of the conditions that are part of, or candidates for, newborn screening. An important aspect of these efforts is the use of a common set of questions and answers that enable researchers to combine datasets to accelerate discoveries. NBSTRN developed these common sets, called Common Data Elements (CDEs), by organizing workgroups of leading clinicians who treat individuals with these conditions.

CDEs are available for the majority of diseases that are routinely screened in newborns, and NBSTRN works to develop CDEs for new conditions that are part of newborn screening pilots. We are excited to report that one of these workgroups has recently completed their work and developed over 1500 CDEs for six lysosomal storage disorders (LSDs). This workgroup, chaired by Priya Kishnani, MD, MB BS and Melissa Wasserstein, MD, is made up of 30 members, and their recommended CDE sets for Niemann Pick, Fabry, Gaucher, Pompe, Krabbe, and MPSI will be available on the LPDR website for use by researchers.

The CDE set specific to Pompe will be included in an upcoming guidance document developed by a newborn screening workgroup of the Clinical and Laboratory Standards Institute (CLSI). The overall mission of the CLSI newborn screening workgroups is to lead, oversee, and expand the portfolio of CLSI guidance to meet the needs of the newborn screening community, and NBSTRN thanks CLSI for the opportunity to contribute to this important effort.



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

rfleming@acmg.net

Spotlight on a Newborn Screening Researcher

November's spotlight researcher is the Associate Dean of Clinical Research Operations at the Columbia University Medical Center, Associate Director of Columbia University's NCRR-funded Clinical Translational Science Award (CTSA), and Associate Clinical Professor in the division of Pediatric Hematology.

Our mystery researcher focuses her research on genetic screening of newborns and children, as well as studying the translation of laboratory science to clinical research and training. With a special interest in hemoglobinopathies, her current research focuses on optimizing treatments for sickle cell disease. She was also a former Fulbright scholar and honorary member of the Society for Maternal-Fetal Medicine.

She served as the Medical Director of the National March of Dimes from 2000-2007, where she supervised the scientific priorities, policies and practices and the educational efforts of the Foundation. She was also part of the HHS Secretary's Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children, where she chaired the Criteria workgroup.

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

IDF SCID Newborn Screening Campaign

Since Severe Combined Immunodeficiency (SCID) has been added to the Advisory Committee's Recommended Uniform Screening Panel (RUSP), many states have implemented screening for SCID, but not every state. The Immune Deficiency Foundation (IDF) wants every state to include SCID on their newborn screening panel and has created awareness materials to provide education and information about this issue. To find these resources please visit IDF's website and make sure to watch the "SCID: A Family's Journey Through Isolation " video.

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