Newborn Screening Translational Research Network

September 2014

NBSTRN NEWSLETTER

Where will you see us next?

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, CA

NBSTRN participates in SCID Study

The NBSTRN participated in the following effort to report data from newborn screening programs conducting screening for severe combined immune deficiency (SCID).

Title Newborn Screening for Severe Combined Immunodeficiency in 11 Screening Programs in the United States

Objectives: To present data from a spectrum of SCID newborn screening programs, establish population-based incidence for SCID and other conditions with T-cell lymphopenia, and document early institution of effective treatments.

Main Outcomes and Measures Infants with SCID and other diagnoses of T-cell lymphopenia were classified. Incidence and, where possible, etiologies were determined. Interventions and survival were tracked.

Results Screening detected 52 cases of typical SCID, leaky SCID, and Omenn syndrome, affecting 1 in 58 000 infants (95% CI, 1/46 000-1/80 000). Survival of SCID-affected infants through their diagnosis and immune reconstitution was 87% (45/52), 92% (45/49) for infants who received transplantation, enzyme replacement, and/or gene therapy. Additional interventions for SCID and non-SCID T-cell lymphopenia included immunoglobulin infusions, preventive antibiotics, and avoidance of live vaccines. Variations in definitions and follow-up practices influenced the rates of detection of non-SCID T-cell lymphopenia.

Conclusions and Relevance Newborn screening in 11 programs in the United States identified SCID in 1 in 58 000 infants, with high survival. The usefulness of detection of non-SCID T-cell lymphopenias by the same screening remains to be determined.

To view the list of authors and the full article (available to *JAMA* subscribers), please visit http://jama.jamanetwork.com/article.aspx?articleid=1896983#Introduction

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Spotlight on a Newborn Screening Researcher

Our Spotlight Researcher of the Month is currently a professor of medicine and pathology at the Mayo College of Medicine as well as director of the immunology laboratory at the Mayo Clinic in Rochester, Minnesota. Her formal education and training is in microbiology and immunology.

Her research interests include clinical laboratory immunology related to immunodeficiencies and laboratory assessment of immune competence and function. She has authored over 25 peer-reviewed papers and 21 abstracts.

Find out who the NBS 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

September is Newborn Screening Awareness Month #NBSMonth. Each year in the U.S. 4.1million newborns are screened for certain genetic conditions at birth and around 12,000 infants are diagnosed with a condition listed on the Newborn Screening (NBS) panel. The NBSTRN is proud to support researchers who are involved in newborn screening research. Our research tools, the Virtual Repository of Dried Blood Spots, (VRDBS) the Longitudinal Pediatric Data Resource (LPDR) and the Region 4 Stork Laboratory Performance Database (R4S) all contribute to a researcher's ability to discover new technology, new treatments and new diseases in newborn screening.

To find out more about NBSTRN tools and resources that aid NBS research you can visit the NBSTRN.org or the NBSTRN Research Tools page.

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