Hyper-IgE syndrome, among others. She has been part of gene therapy development for patients with X-linked SCID who failed bone marrow transplantation and now Artemis-deficient SCID. Noting that early diagnosis with treatment prior to occurrence of infectious complications is the key to successful outcomes for SCID patients, she conceived and developed a test to screen newborns for SCID using PCR quantitation of T cell receptor excision circles (TRECs) in DNA isolated from infant dried blood spots. TREC testing, now part of the standard newborn screening panel in several states, allows infants affected with SCID and other T cell lymphopenic disorders to be detected early and treated.
Background
Newborn screening (NBS) for severe combined immunodeficiency (SCID) using T cell receptor excision circles (TRECs) was initiated in Wisconsin in 2008, and added to the DHHS Secretary's Recommended Uniform Panel of newborn screened diseases in May 2010. Through May, 2013, 15 states and the Navajo Nation had adopted universal SCID NBS, with additional states planning to do so.

Objective
To assemble data reflecting the spectrum and experience of state SCID NBS programs.

Methods
State public health laboratory professionals and state-designated immunologists were invited to submit SCID screening algorithms, overall test performance data and de-identified clinical and laboratory information regarding total cases screened and cases with non-normal results. Numbers of infants with SCID and other diagnoses were classified according to standard criteria, and immunological interventions were recorded.

Results
Ten states and the Navajo Nation participated, representing a total of 3,031,883 infants screened with a TREC test. Diagnoses included 52 cases of SCID disorders, typical SCID, leaky SCID and Omenn syndrome, affecting 1 in 58,000 infants. There were 398 additional cases of T cell lymphopenia, or 1 in 7,700 infants. The infants identified received immune restoring interventions including hematopoietic cell transplantation, thymus transplantation, enzyme replacement therapy and gene therapy, as well as protective interventions including immunoglobulin infusions, antibiotics, transfusion precautions, and live vaccine avoidance.

Conclusions
NBS successfully identified SCID cases early in life, making possible prompt intervention previously shown to optimize survival. The experience of states already performing SCID NBS will facilitate adoption by additional states in the U.S. as well as by other countries.