

New SCID Screening Saves Lives

Newborn Screening Program August 2014 Announcement

Nebraska Program to add Severe Combined Immune Deficiency to the Newborn (Blood-spot) Screening Panel

Effective October 4, 2014 babies whose specimens are received at Nebraska's newborn screening laboratory (PerkinElmer Genetics) will be screened for Severe Combined Immune Deficiency (SCID). Some people may remember the 1970s movie "The Boy in the Bubble" in which a young John Travolta portrayed the life of David Vetter. In real life, David was affected with SCID, and despite treatment died at the age of 12.

Babies born with SCID do not produce T-cells and are highly susceptible to common childhood infections which can result in infant or early childhood death. Undiagnosed, these babies often die by the age of 2.

We have learned much from David Vetter and other patients since then about the treatment of SCID and other related T-cell immune deficiencies. We know that bone-marrow stem cell transplant (BMST) can be very effective in re-constituting these patient's immune systems. We also know that when this accomplished by 3 months of age and prior to the onset of any infections the success rate and long-term survival are dramatically improved.



Nebraska now has a screening test available which is called the TREC assay. Essentially, TRECS are elements of the DNA that are excised when T-cells are going through recombination. Zero or low copy numbers of TRECS found on this test are a surrogate marker for no or low T-cell production and will trigger additional follow

up. The test which has been implemented in about 20 other states has proven to be highly sensitive and specific.

The program also has a recommended protocol for when positive screen results are identified. The goal is to prevent infection until the BMST can be conducted, and to get this done by 3 months of age. When a baby's healthcare provider is notified of a positive result the Newborn Screening Program will provide:

- An action (ACT) sheet recommending next steps for the provider.
- A parent information (FACT) sheet, including links to appropriate website resources that the provider can hand to the parent.
- Connection to the pediatric immunologist on call at the time the result is available, for immediate consultation on further testing and recommendations for ensuring protective isolation of the newborn.

We have also learned from the pilot studies and other states' screening that the screening test for SCID can identify babies at risk for several immune deficiencies (not all) and T-cell lymphopenias. We expect to identify a baby with true SCID about once every 1 to 1.5 years. However, it is likely babies with other immune deficiencies discovered through the differential diagnosis, will also benefit from this screen. The cost of the screen for all 29 conditions will increase by \$5.50. The total charge for a newborn screen will now be \$45.

For more information: www.dhhs.ne.gov/nsp or e-mail: dhhs.newbornscreening@nebraska.gov

