



Severe combined immune deficiency (SCID) is an immune disorder characterized by profound defects in T-lymphocyte and B-lymphocyte function, rendering affected babies susceptible to multiple types of life-threatening bacterial, viral, fungal and opportunistic infections. Early diagnosis of SCID is a **pediatric emergency**, and SCID is **curable** with hematopoietic cell transplantation if detected early, before the onset of severe, disseminated infections. Without immune reconstitution through bone marrow transplantation, enzyme therapy or gene therapy, most babies will die within the first year of life and SCID is uniformly fatal within the first two years of life. Affected babies can also develop severe complications from live viral vaccines (e.g. rotavirus, MMR) and can develop lethal complications if they receive non-irradiated blood transfusions. Early identification of infants affected with SCID, ideally within the first 3.5 months of life, results in improved outcomes following transplant (>90% survival) as well as decreased long-term medical and developmental complications. Infants with SCID generally do not exhibit any characteristic physical findings and typically appear “well” at birth. However, after transplacentally acquired maternal antibodies begins to decline, they become susceptible to infections and may exhibit poor weight gain and failure to thrive. Mutations in at least 13 different genes have been found to lead to the clinical phenotype of SCID.

The need for newborn screening for SCID has been recognized for the last two decades, but was limited by the lack of cost-effective methods for newborn screening. Approximately 7 years ago, significant progress toward implementation of SCID newborn screening was made with development of an assay that could be performed on the dried blood spots (“Guthrie spots”) routinely collected from newborn infants. This assay quantifies the number of T-cell receptor excision circles (TRECs) which are small pieces of circular genetic material that are generated during the T-cell education process in the thymus. This assay not only identifies babies with SCID, but also identifies babies with low T-cell numbers (T-cell lymphopenia) from other causes such as DiGeorge Syndrome and other combined immune deficiencies. Wisconsin was the first state to implement universal newborn screening for SCID in 2008. In May 2010, Secretary of Health and Human Services Kathleen Sebelius approved the recommendation of the Secretary’s Advisory Committee on Heritable Diseases in Children (SACHDC) to add SCID screening to the Recommended Uniform Screening Panel (RUSP) and to add related T-cell deficiencies to the list of secondary targets. One year later, in May 2011, 6 states and 1 territory were screening for SCID using the TREC assay and had identified 14 cases of classic SCID as well as 40 cases of T-cell lymphopenia that were not related to SCID in a total of 961,925 screened newborns. All cases had received appropriate immunologic referral and evaluation and all cases had received appropriate therapy.

Multiple states and territories are now screening for SCID including California, New York, Texas, Massachusetts, Florida, Louisiana, Colorado, Minnesota, Delaware, Puerto Rico with many others beginning pilot programs. Although the true incidence of SCID is not known, following the advent of newborn screening, many experts suggest that is much more common than previously thought (approx. 1/100,000), with the estimated incidence of SCID now approximately 1/40,000, and screening has allowed for increased detection in minority populations that could have been missed previously. Further, states that are screening for SCID estimate significant cost savings; in Wisconsin, five babies with a late diagnosis averaged approximately \$2.2 million per child in hospital costs, only three survived, whereas the hospitalization and treatment for an infant with an early diagnosis was \$250,000. other states have also reported significant savings for their Medicaid programs.

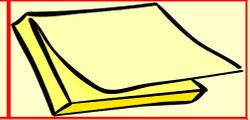
In summary, newborn screening for SCID and T-cell lymphopenia is a cost-effective, life-saving approach to diagnosing a curable disorder in infancy. When detected, SCID results in extended, expensive hospital stays and poor clinical outcomes, as well as significant psychological effects on families due to the unmitigated tragedy of infant loss. Addition of screening for SCID to state newborn screening panels is a rational approach to improve for these patients and families and we are eager to begin screening in Arkansas.

#### **References:**

1. Buckley RH. The long quest for neonatal screening for severe combined immune deficiency. *J Allergy Clin Immunol.* 2012; 129; 597-604
2. Puck JM. Laboratory technology for population-based screening for severe combined immune deficiency in neonates: the winner is T-cell receptor excision circles. *J Allergy Clin Immunol.* 2012; 129: 607-616
3. [www.hrsa.gov/advisorycommittees/mchbadvisory/heritabledisorders/reports/committeesSCIDreport.pdf](http://www.hrsa.gov/advisorycommittees/mchbadvisory/heritabledisorders/reports/committeesSCIDreport.pdf)
4. Myers LA, Patel DD, Puck, JM, Buckley RH. Hematopoietic stem cell transplantation for severe combined immune deficiency in the neonatal period leads to superior thymic output and improved survival. *Blood* 2002; 99; 872-8.
5. [www.aphl.org/conference/proceedings/Documents/2010/2010 APHL Annual Meeting/053Brokopp.pdf](http://www.aphl.org/conference/proceedings/Documents/2010/2010%20APHL%20Annual%20Meeting/053Brokopp.pdf)

*Nursing Notes from Mary Jean:  
Importance of Demographics*

*Volume 1, Issue 1*



Completing the newborn screening form is so vital to the process of the whole screening procedure. Once the circles are filled on the form completely, the baby's information also needs to be completed, correctly and completely. When we receive abnormal newborn screen notices from the lab, we immediately look for the PCP's name for notification; if the name is missing or belongs to the hospital physician, we end up having to call the mother. It is preferred that this is done by the physician. If mom's number is missing, we're in a real quandary. Depending on the nature and severity of the abnormal screen, we

start checking baby's status, type of feeding and first or repeat specimen. Doing this lets us know if the baby is sick and or on TPN, which may affect the MSMS results. The type of feeding is especially important for babies with abnormal GALT results. We ask that mom immediately switch to a soy formula. First or repeat specimen is important. Most often when baby has 2 borderline abnormal results it will mean confirmatory testing will have to be done. If repeat specimen is marked, we immediately know to check results of the first. Even checking the wrong race of a baby may cause problems and be time

consuming in tracking the proper baby down. One form was marked as baby being white and the result of the screen was sickle cell disease. One BB's name was on 2 NBS's, on one he was marked as white, female and mom's name was listed, on the second form he was marked as black male with another mom's name. Thank goodness both screens were normal.

**Please take the time to do it correctly and completely; for babies' sakes.**

Mary Jean Gresham, RN

*Notes from the lab: by Leslie Himstedt*

Collection cards with an expiration date of 5/2013 may be used for collection through May 31st. Any sample collected on these cards after that date will be rejected for use of expired filter paper. It is important that the specimen collection card is **complete and legible**. Please print using block letters. In some cases, the 'baby's last name' section does not provide enough boxes for the entire least name. If this is the case, please continue the name outside the boxes.



It is important that we get a complete last name for proper identification of the baby. Prompt specimen delivery to the lab is critical. Due to the nature of some of the disorders screened for, it is imperative that the samples be received as soon as possible after collection so affected babies may be quickly identified and follow-up testing and treatment started.

"Rules and Regulations Pertaining to Newborn Infant" specify that specimens shall be submitted to the lab within 48 hours of collection.

**Protecting babies through a simple test!**

Leslie

*Check out the newborn screening website at:*

**[www.healthy.arkansas.gov](http://www.healthy.arkansas.gov)**

Click on "N" on A - Z index , then Newborn Screening, you will find helpful resources; and Rules & Regulations in the "Provider" section.

**Next Upcoming State Holiday  
Memorial Day!**



Old State House, original site of the Arkansas Department of Health