Starting in fall 2012 the Iowa Neonatal Metabolic Screening Program (INMSP) will begin screening for severe combined immune deficiency (SCID). SCID is a rare inherited disorder caused by a deficiency or absence of T cells. The symptoms of SCID include recurrent life-threatening infections, failure to thrive, diarrhea and thrush. The average age of diagnosis is approximately 6 months. If not treated, most patients do not survive past one year of age. The incidence of SCID is estimated to be 1 in 50,000 to 1 in 60,000 live births. There are greater than 15 known genetic causes of SCID.

Early detection of this disorder allows for infants to receive necessary life-saving treatment such as intravenous immunoglobulin, antimicrobials, isolation precautions and a bone marrow, peripheral blood stem cell or umbilical cord blood transplant. Testing for SCID involves T-cell receptor excision circle (TREC) analysis.

Although the TREC assay is designed to detect SCID, in some instances it also can detect other T-cell disorders such as 22q deletion syndromes (i.e., DiGeorge anomaly) and certain congenital malformations associated with lymphopenia. Medical consultants will be available to assist with all abnormal TREC assay results.

During this phase of the pilot, SCID results will not be reported routinely. However, the infant’s attending physician will be notified and consultation provided if further testing is indicated. This process will occur exactly as it has in the past for other disorders for which INMSP screens. We anticipate that SCID will be added to the INMSP as a routine component of the test battery within the next year, pending evaluation of pilot data by medical consultants to the Congenital and Inherited Disorders Advisory Committee. Specimen collection will not change from the current procedure. Billing practices remain unchanged and there is no additional increase in the NBS fee associated with the addition of this marker during the pilot testing.

For additional information on SICD newborn screening view the presentation by Dr. Mary Beth Fasano on the development, implementation and science behind Iowa’s SCID newborn screening panel. Dr. Fasano is the Medical Consultant for the State of Iowa’s SCID Newborn Screening Program. View the link at https://www.youtube.com/watch?v=2Sre84FTp_8
The newborn screening dried blood spot card collection process will not change. Once the dried blood spot card is received at the State Hygienic Lab in Ankeny, Iowa, an additional testing procedure, T-cell receptor excision circle (TREC) analysis will be performed. Full-term infants with an abnormally low TREC and preterm infants (< 37 weeks gestation) with a TREC of 0 will be followed up emergently. A member of the Iowa Newborn Screening Program will call the primary care physicians (PCP) immediately upon being informed of the result with further recommendations.

Infants with a presumptive positive SCID result will require hospitalization at the University of Iowa Hospitals and Clinics (UIHC) to be cared for by a team of experts in the evaluation and management of SCID. Infants with a borderline SCID result will need to be evaluated at the PCP clinic on the same day test results are received and a blood sample for second tier testing will need to be sent to UIHC. The same courier system can be used to send your second tier blood sample as dried blood spot cards. Once the samples are received at UIHC flow cytometry will be performed. If second tier test results are abnormal a medical consultant from UIHC will contact the PCP immediately to provide assistance on how to further treat this infant.

During the pilot phase only abnormal TREC results will be reported from the State Hygienic Lab. This pilot phase will be used to evaluate and improve our state-specific cutoff values. If you have any additional questions please do not hesitate to contact the Iowa Newborn Screening Program at 319-384-5097 or 1-800-890-5965.

Dr. Mary Beth Fasano’s presentation at Grand Rounds at the University of Iowa is available here [https://www.youtube.com/watch?v=2Sre84FTp_8](https://www.youtube.com/watch?v=2Sre84FTp_8) and is a good resource for more information.
Questions regarding SCID screening may be directed to the following persons

Newborn screening questions: Iowa Neonatal Metabolic Screening Program  
319-384-5097  
1-866-890-5965  
iowanewbornscreening@healthcare.uiowa.edu

Laboratory questions: Stan Berberich, Ph.D.  
Manager, Newborn Screening Laboratory  
319-335-4500  
stanton-berberich@uiowa.edu  
Mike Ramirez, B.S.  
Supervisor, Newborn Screening Laboratory  
515-725-1630  
michael-ramirez@uiowa.edu

Follow-up and treatment questions: Mary Beth Fasano, MD, MSPH  
Pediatric Immunologist  
Medical Consultant for SCID  
319-384-5097  
iowanewbornscreening@healthcare.uiowa.edu

Administrative questions: Kim Piper, RN, BS, CPH, CPHG  
State Coordinator for Genetic Services  
515-720-4925  
kimberly.piper@idph.iowa.gov