

NEWBORN SCREENING NEWSLETTER

JULY 2013



Severe Combined Immunodeficiency (SCID) Screening to begin on 7/29/2013

Ohio's newborn screening program expands on July 29, 2013 to include screening for Severe Combined Immunodeficiency (SCID) and conditions associated with Severe T-cell Lymphopenia. This program addition was recommended by the U. S. Department of Health and Human Services' Discretionary Advisory Committee on Heritable Disorders in Newborns and Children and the Ohio Department of Health Newborn Screening Advisory Council. This additional test does **not** require additional blood to be collected.

The newborn screening kit fee will increase on the same day from \$55.16 to \$63.61 to cover the cost of the additional test and program change.

The Screening Process for SCID

What is it?

Severe combined immunodeficiency (SCID) includes a group of rare, serious, and potentially fatal inherited immune disorders in which T lymphocytes fail to develop and B lymphocytes are either absent or compromised. Impairment of both B and T cells lead to the term combined immunodeficiency. Untreated patients develop life-threatening infections due to bacteria, viruses, and fungi. The screening test for SCID is TRECs (T cell receptor excision circles), which are a by-product of normal T cell development. This test identifies SCID as well as certain related conditions with very low T cell number or function.

Clinical Features

Infants with SCID appear to be normal at birth, but without early treatment/intervention, these infants cannot survive. In most instances, they will have a low lymphocyte count, absent thymic shadow on chest radiograph, poor growth and will be at risk for opportunistic infections.

Treatment

The definitive treatment for most children diagnosed with SCID is a bone marrow transplant. For some children, gene therapy may be an alternative. Infants diagnosed with SCID should be placed on replacement gamma globulin therapy, and they may also require prophylactic antibiotics. They should avoid public places in order to limit exposure to infections. They should not receive any live viral vaccines. If transfusions are required, they should only receive leukocyte reduced, irradiated blood products that are negative for cytomegalovirus (CMV).

Screening Results

Screening results will be reported in one of 3 ways: (1) Low Risk – TRECs are Within Normal Limits, (2) Moderate Risk – TRECs are Decreased, (3) Elevated Risk – TRECs are Absent.

Follow-up of At-Risk Screening Results

The ODH Lab will call the Primary Care Provider (PCP) to report results that indicate an Elevated or Moderate risk and to explain the recommended clinical follow-up. The PCP will also receive a faxed letter specifying the results and appropriate follow-up, as well as a referral list for immunologists in Ohio. The PCP should communicate the results and follow-up plan to the family immediately. The PCP should stress the need to limit the baby's exposure to infections while the diagnostic work-up is underway.