

# SCID Test Fact Sheet for Physicians

## GENERAL INFORMATION:

T cells are lymphocytes that are critically important in immunity to infection. The State of Wisconsin screens all newborns for a deficiency of T cells using the “TREC assay.” TREC stands for “T cell receptor excision circles.” TRECs are pieces of DNA that are formed during T cell development and can be measured in an infant’s blood. TRECs are surrogate markers for the number of T cells in the blood.

A variety of gene mutations can lead to a low number of T cells. Depending on the T cell count, the immunodeficiency can be quite serious. The most serious immune deficiency is severe combined immune deficiency (SCID). Babies born with SCID are at high risk for serious infections and will likely die within the first year of life unless identified and given a bone marrow transplant. Less severe deficiencies of T cells need monitoring by immune specialists.

TRECs are quantified on DNA extracted from the dried blood spot on the newborn screening card using the polymerase chain reaction (PCR) method. A measurement below 40 TRECS/microliter of blood is considered a screening positive, warranting a more definitive test to quantify the number of T cells in the blood. Screening positive results are communicated to the primary care physician with instructions to arrange for immediate testing. The screening positive results are also reported on the comprehensive newborn screening report that is sent to the submitter of the specimen.

## WHAT TO DO WHEN YOUR PATIENT HAS AN ABNORMAL SCID TEST:

Babies with screening positive SCID tests at birth urgently need an additional blood test to confirm the presence or absence of an immune deficiency. All abnormal test results are communicated immediately to one of the immunologists listed below. They will contact you to arrange for your patient to have confirmatory blood testing. As your families will also have questions about this process, a fact sheet for parents (P-00855) is available at: <http://www.dhs.wisconsin.gov/publications/P0/P00855.pdf>

Immunologists are available on call 24/7 to discuss the abnormal test results in more detail. (See below):

Name	Contact Number	Institution
John Routes, MD	414-907-3644 (pager)	Children’s Hospital of Wisconsin, Milwaukee
Christine Seroogy, MD	608-263-2652	University of Wisconsin American Family Children’s Hospital, Madison

The content of this fact sheet has been reviewed by the Newborn Screening Advisory Subcommittee.



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