

Fact Sheet for Providers:

New York Becomes the 4th State to Offer Newborn Screening for Severe Combined Immunodeficiency

When was severe combined immunodeficiency (SCID) added to the newborn screening panel?

On September 29, 2010, New York State added severe combined immunodeficiency (SCID) to its newborn screening panel, allowing affected infants to receive treatment sooner.

Why was SCID added to the newborn screening panel?

The Secretary of the U.S. Department of Health and Human Services Advisory Committee on Heritable Disorders in Newborns and Children directed an evidence review regarding newborn screening for SCID and the findings were published in January 2009. After receiving a letter of recommendation from the Committee, Kathleen Sebelius, Secretary of Health and Human Services, recommended that SCID be included in the uniform newborn screening panel on January 21, 2010.

What is SCID?

SCID is a rare genetic disorder caused by a deficiency or absence of functional T cells. The symptoms of SCID include recurrent 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 1/70,000 to 1/100,000 live births. There are greater than 15 known genetic causes of SCID.

What are the benefits of adding SCID to the newborn screening panel?

Newborn screening will allow infants to be diagnosed and treated sooner. Infants with SCID, who have a sibling with the condition have received treatment earlier than average. Experience with these infants has provided evidence that early diagnosis and treatment likely leads to better outcomes and fewer complications.

How will the Newborn Screening Program screen for SCID?

The NYS Newborn Screening Program will screen for SCID by evaluating the number of T cell receptor excision circles (TRECs) in the dried blood spots currently collected from newborns. TREC analysis has been researched extensively as a reliable and sensitive method. TRECs are a piece of DNA produced during the formation of T cells in the thymus. Although this testing is DNA-based, TREC analysis is not a test for gene mutations. TRECs may also be low in infants with non-SCID-related causes of T cell lymphocytopenia, who will also require evaluation and management.

What will happen if an infant screens positive for SCID?

Infants who screen positive will be referred to a pediatric immunologist or infectious disease specialist at a Specialty Treatment Center for a diagnostic evaluation. There is a pediatric immunologist or infectious disease specialist at eight Specialty Treatment Centers across the state, and infants requiring follow-up will be referred to the provider closest to the family.

What will the initial diagnostic evaluation consist of?

The initial diagnostic evaluation will include a CBC and flow cytometry and if this testing is normal, then further follow-up will not be needed. For infants with confirmed SCID, the pediatric immunologist or infectious disease specialist will continue to follow the infant after the completion of the diagnostic evaluation. They will make recommendations for medical management and treatment. The infants will receive immunoglobulin therapy and antibiotics and they will also be placed in isolation. The parents will be offered hematopoietic stem cell transplant, which has been shown to restore immune function.

**Newborn Screening Program staff can be contacted at
518-473-7552, Monday through Friday, 8 am until 5 pm.**



**State of New York
Department of Health**

