

## **Severe Combined Immunodeficiency Fact Sheet for Physicians**

### **What is SCID?**

SCID is a rare inherited disorder caused by a deficiency or absence of T cells. The symptoms of SCID include recurrent infections, failure to thrive, diarrhea and thrush. 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, however since initiating newborn screening, the incidence has been found to be higher. There are multiple 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. Early diagnosis and treatment saves lives and improves outcomes.

### **How will the Newborn Screening Program screen for SCID?**

SCID screening involves evaluating the number of T cell receptor excision circles (TRECs) in the dried blood spots currently collected from newborns. 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 lymphopenia, who will also require evaluation and management.

### **What will happen if an infant screens positive for SCID?**

Infants whose newborn screen is positive will need to have a clinical evaluation and blood sample drawn for CBC with differential and flow cytometry to determine the extent of the T cell lymphopenia. This evaluation will be performed by the Immunologists at Arkansas Children's Hospital. Those infants requiring stem cell or bone marrow transplant will be referred to a specialized transplant center.

### **What will the initial diagnostic evaluation consist of?**

The initial diagnostic evaluation will include a CBC with differential and flow cytometry as well as additional assessment of B cell and T cell immunity. For infants with confirmed SCID, the pediatric immunologist will make further recommendations for management and treatment which may include hospitalization, isolation, immunoglobulin therapy and antimicrobial agents to decrease the risk of infection. The parents will be offered hematopoietic stem cell transplant at a specialized transplant center. Early transplant has been shown to restore immune function and be curative for SCID.