KANSAS DEPARTMENT OF HEALTH AND ENVIRONMENT

NEWBORN SCREENING ACT SHEET

SCREEN FOR: T CELL RECEPTOR EXCISION CIRCLES (TRECs)

CONDITION: SEVERE COMBINED IMMUNODEFICIENCY (SCID) AND OTHER CONDITIONS ASSOCIATED WITH T CELL LYMPHOPENIAS

METABOLIC DESCRIPTION: Severe Combined Immune Deficiency (SCID) is a group of rare but potentially life-threatening, genetic disorders in which T lymphocytes fail to develop in addition to potential abnormal levels of B lymphocytes and NK cells. This "combined" impairment leads to life-threatening bacterial, viral, and fungal infections. The T cell receptor excision circles (TRECs) test is a screening test for a byproduct of normal T cell receptors. The TRECs test can identify SCID along with other disorders with low T cells such as DiGeorge syndrome and ataxia telangiectasia at birth.

MEDICAL EMERGENCY - ACTION TO BE TAKEN **IMMEDIATELY:**

- Contact the family to inform them of the newborn screening result as additional tests are needed to determine if there is actually an immune deficiency.
- If there is any evidence of infection, refer to Children's Mercy or the University of Kansas Medical Center emergently for further evaluation and treatment.
- If the infant requires blood products, ensure they are leukoreduced, irradiated, and CMV negative.
- **DO NOT** give any live attenuated viral vaccines, which could cause serious illness. Avoid live viral vaccines in family members who have contact with the child.
- Infants may not be able to be breastfeed depending on the CMV status of the mother.
- Consult pediatric immunology immediately.
- Provide the family with information about SCID and T cell lymphopenia.
- Report confirmatory findings to the newborn screening program.

CONFIRMATION OF DIAGNOSIS: Confirmatory testing may include repeating the newborn screening, particularly in premature infants, CBC with differential, absolute lymphocyte counts to determine presence or absence of particular lymphocyte (T, B, NK) subsets, lymphocyte functional assays, and genetic testing. The immunologist may order these tests and coordinate further care, and offer genetic counseling.

CLINICAL EXPECTATIONS: It is possible that the infant may need immunoglobulin infusions and prophylactic antibiotics to protect against infections. Patients may present with diarrhea, thrush, failure to thrive, serious infections (pneumonia, meningitis, and/or sepsis), common infections (bronchitis or otitis), and opportunistic infections often begin by 2-4 months of life. Bone marrow hematopoietic cell transplantation may be curative with greater success if done before 3 months of life. Enzyme replacement and gene therapy may be available for some types of SCID. Specific gene diagnosis is important for long term management and genetic counseling.

ADDITIONAL INFORMATION:

Genetics Home Reference	http://ghr.nlm.nih.gov/condition/x-linked-severe-combined-immunodeficiency
SCID.net	http://www.scid.net/
Immune Deficiency Foundation	http://primaryimmune.org/
NMDP:	https://bethematch.org/for-patients-and-families/learning-about-your-disease/severe-combined-immunodeficiency/

PEDIATRIC IMMUNOLOGY SPECIALISTS:

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